THE HUMAN GENETIC MUTANT CELL REPOSITORY

List of Genetic Variants, Chromosomal Aberrations

and Normal Cell Cultures

Submitted to the Repository

Fourth Edition

October 1977

INSTITUTE FOR MEDICAL RESEARCH

Copewood and Davis Streets

Camden, New Jersey 08103

609-966-7377

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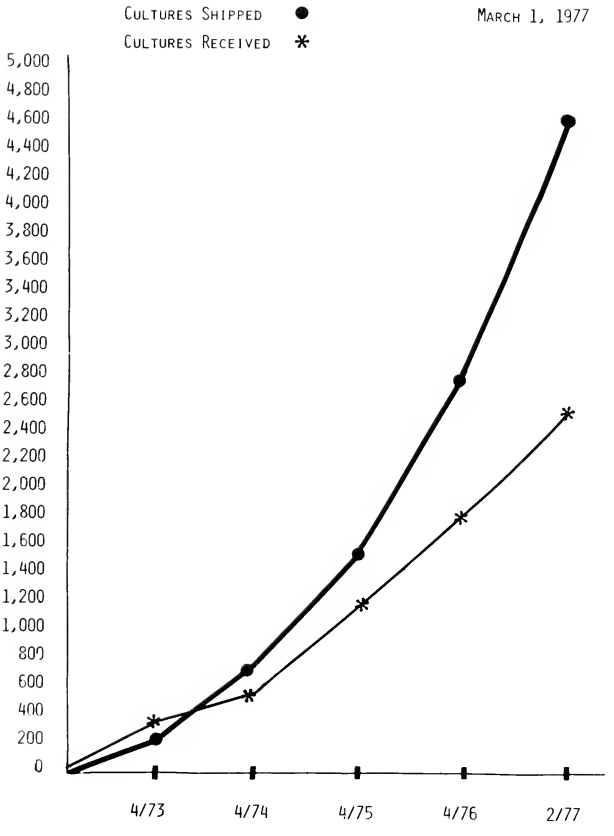
Preface to the Fourth Edition

Acquisition of cell cultures by the Repository and shipment of cell cultures to investigators both continue to increase at an accelerating rate as shown on the graph which follows this preface. In addition to many new cell cultures and supplementary data on others, the present edition contains new features. McKusick's numbers, where applicable, are inserted immediately following the name of each disease. A bracket is placed around cell cultures from members of a family group. HL-A antigens have been determined on many of the lymphocyte cell cultures and this data is recorded in the table on pages 161-163. Eleven cultures are available from both this Repository and from the American Type Culture Collection in Rockville, Maryland under a different numerical designation. To avoid confusion we have listed the ATCC designation under "Remarks".

Camden, N.J., August 1977 Lewis L. Coriell, M.D, Ph.D.

Arthur E. Greene, D.Sc.

THE HUMAN GENETIC MUTANT CELL REPOSITORY



I. INTRODUCTION

The Human Genetic Mutant Cell Repository established in 1972 by the National Institute of General Medical Sciences at the Institute for Medical Research, Camden, New Jersey, contains low passage skin fibroblast, lymphoblast, amniotic fluid cell, and a few animal cell cultures stored in liquid nitrogen from hereditary diseases including those with biochemical and chromosome abnormalities and normal controls. Cell cultures stored in the Repository are verified for freedom from contamination, species of origin, karyotype, viability and expression of the biochemical or chromosomal defect. These cells are available to organizations or individuals engaged in health-related research or health delivery concerning early diagnosis, prevention, treatment, counseling and research for control of some of the 2,000 or more inherited diseases that afflict human beings. The problems in many of these diseases are gene abnormalities which alter one or more chemical steps in the normal metabolic sequence of cells. The application of techniques for detailed study of these cellular chemical processes in cell culture is leading to a wealth of information about human genetics and improvement in the prevention and treatment of genetic diseases and probably many other diseases in which genetic factors are not yet recognized to play a significant role. The Repository program has been developed by the National Institute of General Medical Sciences with the help of a scientific advisory committee which periodically reviews the progress of the

collection and provides advice on the acquisition of cell lines.

Cell cultures are listed alphabetically under cell type and biochemical or morphological abnormalities including the genetic mutant (GM) repository number, tissue, passage number, culture medium, submitter, age, sex, race, genetic status, verification and remarks.

Appreciation is expressed to the many investigators who help the NIGMS Advisory Committee and the Repository staff select representative subjects and help validate the genetic defects by providing clinical and laboratory data or assays on the cell culture.

Interested investigators are invited to utilize the Repository as a source of genetic mutant cell cultures.

Lewis L. Coriell, M.D., Ph.D.

Arthur E. Greene, D. Sc.

II. PROCEDURES FOR ORDERING GENETIC MUTANT CULTURES

1. REQUIREMENTS

The cells are distributed only to qualified professional persons who are associated with recognized research, medical, educational or industrial organizations engaged in health-related research or health delivery. Before fibroblast cells can be shipped, the assurance form (Par. 10) must be signed and returned to the Repository. This is adequate for most cell cultures in the Repository. However, lymphoblast or virus transformed cell cultures require prior agreement to observe the Minimum Safety Guidelines (Page 12) and return of the signed Agreement on Lymphoid and Virus Transformed Cells (Par. 11). These forms may be obtained by writing to the Repository, or may be photocopied.

2. PROCEDURE

Requests for cell cultures must be submitted on institutional purchase forms including purchase order numbers. Purchasing agents should indicate the name of the investigator on such orders. Telephone orders will be accepted when accompanied by a purchase order number, and should be confirmed within several days by letter. Each culture requested should include the Genetic Mutant Repository number (GM number), and the diagnosis.

3. ADDRESS

All requisitions should be addressed to:

Dr. Arthur E. Greene

The Human Genetic Mutant Cell Repository

Institute for Medical Research

Copewood and Davis Streets

Camden, New Jersey 08103

Telephone 609-966-7377

4. FEES

The fee for each 25 cm^2 flask of cells is $20.00 to non-profit institutions, but is reduced one dollar per culture up to 5 orders received simultaneously i.e. $19.00 each for an order of 2 cultures, $16.00 each for 5 or more. Frozen ampules are not shipped, because experience has shown much better success in shipping freshly revived flask cultures. Mass production of cell cultures is not a function of the Repository, (See Appendix E). The Repository is designed to provide only seed cultures. It is suggested that the recipient store aliquots of early passages in liquid nitrogen as insurance against contamination, accidents, artifacts associated with aging, and loss of the culture.

5. SHIPPING CHARGES

The shipping charges are prepaid and will be added to the invoice at the time of billing.

6. HOW SHIPPED

All cell cultures are grown and frozen in antibiotic-free media to aid in detection and prevention of contamination. Cell cultures in the Repository have been tested and found free of mycoplasma, bacteria, molds and fungi during characterization, at the time of frozen storage, and after recovery from liquid nitrogen. When an order is received, a frozen ampule is usually recovered from liquid nitrogen on the following Thursday and the medium is changed on Friday. The following Monday, the culture is inspected, the medium is removed and the T25 flask is filled with fresh medium, packed and shipped, usually by air mail special delivery. The flask is enclosed within two watertight plastic envelopes within a styrofoam box in a cardboard mailer to prevent leakage, overheating or freezing during shipment. A return postcard is enclosed. Please return the postcard with notations about the condition on arrival. Pertinent suggestions to improve the effectiveness of the Repository are appreciated.

7. DESCRIPTIVE DATA

Each shipment contains descriptive data about the cell culture, suggested directions for cell growth and pertinent references when available. Upon receipt cell cultures should be placed in the incubator at 37°C for a few hours or overnight to permit recovery from damage which may have occurred during shipment.

8. REQUEST FOR CITATION AND REPRINTS

It will be greatly appreciated and will make the cell collection more valuable if the source and Repository number is cited in publications in which cell cultures from the Human Genetic Mutant Cell Repository are used. The Repository would appreciate receiving a reprint of such publications.

9. PUBLICATION OF DESCRIPTIVE ARTICLES ON CELL CULTURES STORED IN THE REPOSITORY

Concise descriptions of some of the cultures stored in the Repository are published after characterization in Cytogenetics and Cell Genetics under the name of the individual originally submitting the culture or biopsy. These descriptions include brief clinical histories, assay methods when applicable, family pedigrees, full or partial karyotypes in the case of chromosomal aberrations, and references to previous work involving the cell lines. For a listing of these concise papers, see Appendix C, page 153.

10. ASSURANCE FORM

Before cells can be shipped from the Genetic Mutant Cell Repository the recipient institution must submit a purchase order number and agree to the limitations listed below. This agreement must be renewed annually. Please sign and return originial to the Institute for Medical Research, Copewood Street, Camden, New Jersey 08103.

ASSURANCE

As purchaser of cell cultures from the Genetic Mutant Cell Repository we agree that such cells, their progeny, or derivatives will not be used in human experimentation. It is further agreed that if such use is planned, the purchaser will first obtain prior written approval of the Project Officer, NO1-GM-6-2119, National Institute of General Medical Sciences.

It is further agreed that cell cultures obtained from the Genetic Mutant Cell Repository will not be resold, but they may be replicated by a third party for the original purchaser. The third party shall not be allowed to resell other than to the original purchaser furnishing the material for replication and the third party shall not use such cells for human experimentation.

Name of Institution	Name of Principal Investigator	Date
Signature of Authorized Official	Signature of Principal Investigator	Date

11. AGREEMENT ON LYMPHOID AND VIRUS TRANSFORMED CELLS

This document must be appropriately completed and exchanged before lymphoid and virus transformed cell lines can be transferred from the Human Genetic Mutant Cell Repository to your laboratory.

AGREEMENT

The potential hazardous nature of these human cell lines is unknown. However, in view of this lack of knowledge, it is appropriate that the scientific community be made aware that a potential hazard may exist. Therefore, we recommend that the enclosed guidelines for laboratory procedure be adhered to in the handling of lymphoid and virus transformed cell lines. They are also recommended for handling all cell cultures.

It is understood that you will not further distribute cell lines sent to you to laboratories not under your direct supervision. It is further agreed that all cultures obtained from the Human Genetic Mutant Cell Repository may only be replicated by a third party if the third party also executes an Agreement on Lymphoid and Virus Transformed Cells agreeing to follow the Minimum Safety Guidelines.

As purchaser of cell cultures from the Human Genetic Mutant Cell Repository we agree that such cells, their progeny, or derivatives will not be used in human experimentation. It is

further agreed that if such use is planned, the purchaser will first obtain prior written approval of the Project Officer, NO1-GM-6-2119, National Institute of General Medical Sciences.

This agreement, when appropriately executed, will be on file and will qualify you to receive lymphoid and virus transformed cell lines from the Human Genetic Mutant Cell Repository.

AGREEMENT ON LYMPHOID AND VIRUS TRANSFORMED CELLS

We the signatories below, have read and understand this document and agree that in the handling of cell lines furnished, we will adhere to the procedures and recommendations outlined in the foregoing and the attached "Minimum Safety Guidelines Recommended for Working with Lymphoid and Virus Transformed Human Cell Lines."

A. For the recipient laboratory:

Investigator: Signature _____ Date_____

Title _____

Head of Laboratory or Immediate Supervisor: Signature _____ Date_____

Title _____

Address _____

B. For Human Genetic Mutant Cell Repository:

Signature _____ Date_____

Title _____

III. HUMAN GENETIC MUTANT CELL REPOSITORY MINIMUM SAFETY GUIDELINES RECOMMENDED FOR WORKING WITH LYMPHOID AND VIRUS TRANSFORMED HUMAN CELL LINES*

A. Supervision

1. Administrative Responsibilities

a. Responsibility of Management

Management should establish a biohazards committee to institute and enforce a health and safety policy which includes a specific safety program for work involving human cell lines. The program should meet applicable federal, state and local regulations and include safety training, maintenance of accident records, and provisions for emergency treatment.

b. Responsibility of the Principal Investigator

The principal investigator is responsible for the preparation of safety protocols for the research program under his direction. The protocols should include appropriate procedures for use, storage, decontamination, disposal and emergency treatment. The protocols should be approved by the biohazards

*These guidelines were developed on February 11, 1974 at a special meeting at the National Institutes of Health attended by representatives from the Office of Biohazards of NCI, NIAID, NIGMS and the Advisory Committee to the Human Genetic Mutant Cell Repository.

committee and discussed with the research staff before starting the research program.

2. Medical Surveillance and Screening

a. Physical Examinations

Appropriate pre-employment and periodic medical examinations are desirable for persons working with human cell lines.

b. Work Restrictions

Persons having reduced immunologic competency should be restricted from working with these human cell lines.

c. Serum Collection

Serum should be collected at the time of the pre-employment physical to establish a baseline reference. Serum should be re-collected and stored annually. Serum should be collected immediately after accidental injection or ingestion and at an appropriate interval thereafter. For those individuals exposed to long term lymphoid lines or their derivatives, anti-EB virus titers should be obtained on the collected sera.

3. Laboratory Access

Access to the cell culture area should be restricted to persons directly working with the cell lines, or by specific authorization by the principle investigator or director of the laboratory.

B. Personnel Practices

1. Pipetting

Mechanical pipetting aids rather than mouth pipetting should be used for all pipetting procedures.

2. Eating, Drinking and Smoking

Eating, drinking and smoking should not be allowed in the same areas where cell lines are under study.

3. Protective Clothing

It is recognized that the criteria for protective clothing may vary according to the physical situation of the laboratory and the agents handled. Ideally, adequate protective clothing such as a fully fastened laboratory coat should be worn. This clothing should not be worn outside the work area once the work area has been entered.

C. Physical Control Practices (Recommended for all cell lines, but required for long term lymphoid lines and their derivatives).

1. Ventilated Safety Cabinets or Hoods

Ventilated safety cabinets and hoods and other safety apparatus should be employed and should be tested at least annually to certify correct containment and operation. A list of specifications for satisfactory hoods and instructional materials may be obtained from the Office of Biohazards, NCI.

2. Housekeeping

Appropriate housekeeping procedures which suppress the

formation of aerosols should be used. Working surfaces should be wiped down with an appropriate disinfectant before and after work with each cell culture and at the end of the working day.

3. Decontamination and Disposal

Contaminated glassware and similar materials should be appropriately decontaminated or stored for decontamination before removal from the work area for recycling or disposal. Liquid wastes should be decontaminated either chemically or by heat, before being discharged to the community sanitary sewer system.

4. Protection of Vacuum Lines

Vacuum services, if used, should be protected with disposable absolute air filters and liquid traps. The effluent should be collected in liquid traps containing concentrated disinfectant.

References

Biohazards in Biological Research, ed. A. Hellman, M.N. Oxman and P. Pollack. Cold Spring Harbor Laboratory, New York (1973).

National Cancer Institute Specification: General Purpose Clean Air Biological Safety Cabinet.

IV. SUBMISSION OF SPECIMENS TO THE REPOSITORY

Biopsies are preferred to established cell cultures because they can be processed and stored in lower passage. When submitting a specimen to the Repository, the biopsy should be placed in a 25 cm^2 tissue culture flask or screw top vial with culture medium containing 100 units/ml of penicillin and 100 mcg/ml of streptomycin. Tape the top or cap of the flask very securely to prevent leakage. Package the biopsy flask in a container so that it will not be broken in transit. Mark on the outside of the package that it should be kept at room temperature and not refrigerated, frozen or overheated.

Early passage cultures may be submitted when a biopsy cannot be obtained from the patient. The culture flask (25 cm^2 preferably) should be filled to the top with culture medium and shipped as described above. Mail biopsy or culture the same day by air mail special delivery to:

Dr. Arthur E. Greene
Human Genetic Mutant Cell Repository
Institute for Medical Research
Copewood and Davis Streets
Camden, New Jersey 08103
Phone 609-966-7377

Before a biopsy or cell culture can be processed for the

Repository, documented proof of the diagnosis must be provided on a submission sheet which is available from the Institute for Medical Research, or may be photocopied by the submitter from pages 18-22. Without this information the culture cannot be coded into the information retrieval system or be of value to users of the Repository; and to conserve effort and expense the Repository staff are instructed not to process a specimen unless documentation of diagnosis is provided with the specimen. Submitters are therefore requested to fill in all applicable blanks on the submission sheets. Please provide a family genealogy if appropriate.

No biopsies or cell cultures submitted to the Human Mutant Cell Repository are to be obtained from a live fetus, defined by the presence of pulse, circulation and other vital signs.

SUBMISSION SHEET

Please Complete Every Item that Applies.

TO BE FILLED IN BY IMR

Date Received ___/___/___ (Mo. Day Year)

GM #_____

Contamination ? ____ ____

Failure ? ____ ____

SPECIES: [1] HUMAN (Go to Item 1) [2] OTHER_____ (Specify) (Go to Item 8)

PATIENT

1. Initials: ___ ___

2A. Date of Birth ___/___/___ (Mo. Day Year) or Age: _____ (Circle One) Days Wks. Mos. Yrs.

2B. Gestational Age if Amniotic Fluid or Fetus: ___ ___ Wks. (Circle One)

3. Sex: [1] Male [2] Female [3] Ambiguous [4] Not Recorded

4. A. Race: [1] White [2] Black [3] Oriental [4] Other _____ (Specify)

B. Ethnic Background if Relevant to the Disorder:
(Especially Useful for Inborn Errors of Metabolism)
_____

5. Clinical Phenotype: [1] Clinically Normal [2] Clinically Affected [3] No Information

6. Clinical Manifestations/Diagnosis: (Please List)
A._____
B._____
C._____
D._____
E._____

7. Source(s) of Clinical Information: (Check One or More)
[1] Personal Examination [5] Autopsy Records
[2] Hospital Records [6] Private Physician
[3] Genetic Clinic Records [7] Other _____ (Specify)
[4] Specialist's Report

CULTURE/BIOPSY

8. Local Culture/Lab/Biopsy Identification Number: _____

9. Type of Sample: [1] Culture [2] Biopsy [3] Blood

Passages When Submitted ____

Date of Origin ___/___/___ (Mo. Day Year)

Date Submitted ___/___/___ (Mo. Day Year)

Date Obtained ___/___/___ (Mo. Day Year)

10. Tissue of Origin [1] Peripheral Blood [2] Bone Marrow [3] Amniotic Fluid [4] Skin [5] Other_____ Specify

CULTURE/BIOPSY

11. Culture Medium in which Submitted:

[1] McCoys ___%
[2] Eagle-Hanks' ___%
[3] Eagle-Earle ___%
[4] Ham's F-12 ___%
[5] Ham's F-10 ___%
[8] Other _____ (Specify)

[6] RPMI-1640 ___%
[7] Fetal Calf Serum ___%
[1] Inactivated
[2] Uninactivated

12. Addition of Following to Culture?

Antibacterial Agents?
[1] No
[2] Yes

[1] Penicillin ___%
[2] Streptomycin ___%
[3] Gentamycin ___%
[4] Other ___%
_____ (Specify)

Antifungal (Yeast)?
[1] No
[2] Yes

[1] Fungizone
[2] Mycostatin
[3] Other_____

Other Additives?
[1] No
[2] Yes (List)

_____
_____
_____

13. Special Instruction(s) on Handling or Freezing of Culture/Biopsy?
[1] No [2] Yes _____ (Specify)

PREVIOUS CHROMOSOME STUDIES

14. Have Chromosome Studies ever been Done on this Individual?
[1] Information Not Available (Go to Item 22)
[2] No (Go to Item 22)
[3] No, but Studies are Underway (Go to Item 22)
[4] Yes
[1] On Cells from this Culture
[2] On Cells from Other Tissue/Samples

15. General Karyotype Results: [1] Normal
[2] Abnormal
[1] Balanced
[2] Unbalanced
[3] Not Certain

16. Sex Chromosomal Complement: (Check One or More)
[1] XX [2] XO [3] XXX
[4] XY [5] XXY [6] XYY [7] XXXY
[8] Other _____ (Specify)

17. Autosomal Mosaicism?
[1] Detected (If so, # and % Sub Lines: _____
_____
[2] Not Detected
[3] Suspected

PREVIOUS CHROMOSOME STUDIES

18. Breakage/Somatic Rearrangements?
[1] Data Not Available
2 Data Available
[1] Breaks Observed
[2] Somatic Rearrangements Observed
[3] Neither Observed

19. Current International Nomenclature: _____

_____

_____

20. The Above Karyotype is Based on [1] Banded or [2] Unbanded Technique(s).

21. If Banded, what Staining Method(s) was Used:

_____

_____

PREVIOUS BIOCHEMICAL STUDIES

22. Have Biochemical Studies been Done on this Individual
[1] No (Go to Item 29)
[2] Yes
[1] On Cells from this Culture (Go to Item 23)
[2] On Other Cultures or Tissues from Same Patient (Go to Item 28)

23. Specific Defect Detected or Investigated: _____

_____

24. Assay Method Used: _____

_____

25. Assay Level in this Cell Culture: _____

26. Assay Level in Normal Control Cell Cultures _____

27. Conclusion Based on Biochemical Assay:
[1] Normal [2] Abnormal [3] Inconclusive
[1] Consistent with Heterozygosity
[2] Consistent with Homozygosity
[3] Genotype Uncertain
[4] Not Applicable

28. Significant Laboratory Data on Other Cell Cultures or Tissues from this Individual:

Tissue	Assay	Level	Conclusion (See Item 27)

29. Is Pedigree Available? [1] No [2] Yes

FAMILY HISTORY

30.

Relative	Chrom. Studies Done?	Biochem. Studies Done?	Clinical Lab Studies Done?	Cultures Available?	Banked in Camden?
Father					
Mother					
Siblings					
Offspring					
Other (Specify)					

REFERENCES/COMMENTS/ENCLOSURES

31. Has this Patient/Family been Reported in Literature?
[1] No [2] Yes _____
Specify Reference

32. Has a Sample from this Patient been Previously Stored in Camden?
[1] No [2] Yes, GM #_____.

33. Any Additional Comments/Observations/References/Lab Studies on Culture, Patient, Family Etc.?

_____

_____

_____

Enclosures: [1] Pedigree [2] Clinical History/Description of Patient and/or Family
[3] Karyotype [4] Laboratory Data on Patient/Family
[5] Reprint of Reference [6] Other _____
(Specify)

To encourage storage of unusual cell cultures in the Repository, provision has been made for delayed release to other investigators if the contributor so desires. Please check your preference: a) release culture to anyone requesting it_____, b) release only to contributor during the first year_____. At the conclusion of 1 year the cell culture will be listed in the next printing of the catalog and made available to other investigators unless additional time is specifically requested.

I hereby grant permission for these cells to be stored in a bank of genetic mutant cell cultures and the progeny cells distributed to qualified investigators. Appropriate consent was obtained from the patient from whom the cells were originally obtained, or can be reasonably inferred, for use of these cells for diagnosis, research, teaching or therapy.

No biopsies or cell cultures submitted to the Human Mutant Cell Repository are to be obtained from a live fetus, defined by the presence of pulse, circulation and other vital signs.

Date _____ Submitter _____
(Signature)

Address _____

_____

Telephone Number _____

Mail completed form with, or preferably preceeding shipment of cell cultures, to: Dr. Arthur Greene, Institute for Medical Research, Copewood and Davis Streets, Camden, New Jersey 08103.
Phone: 609-966-7377.

EXPLANATION OF THE CODE INTERPRETING RECORDED

DATA IN THE COLUMNS OF THE CATALOG

COLUMN

GM #: The GM number of the cell culture, refers to the number assigned to the culture when it was received at the Human Genetic Mutant Cell Repository. Fibroblast cultures are all originated from skin unless noted in the Remarks column. Lymphocyte cultures are all established from peripheral blood. McKusick's number is inserted following the name of each disease.*

Passage #: The number of serial in vitro transfers of the cell culture as stored in liquid nitrogen.

Culture Medium: For code see appendix A, page 131.

Submitter Code: A number which identifies the investigator who submitted the biopsy or culture to the Repository. For code see appendix B, page 133.

Age: Age of the donor is expressed in years, or when appropriate in months (mo.), weeks (wk.) or days (da.). F indicates fetus.

Sex: M or F

Race: W-Caucasian; B-black; O-Oriental; I-Indian (India); P-Puerto Rican. Space left empty if race unknown.

*McKusick, Victor. Mendelian Inheritance in Man 4th Edition. John's Hopkins Press, Baltimore, London. 1975.

Genetic Status:	+	Normal gene
	-	Affected gene
	y	Hemizygous for X-linked trait
	?	At risk for autosomal trait, genetic status not determined
	(0)	Carrier for trait as determined by pedigree or clinical diagnosis

The McKusick* number immediately following the name of the disease will indicate the dominant, recessive, or X-linked nature of the disease. The first digit is one for a dominant disease (e.g. Huntington Chorea - 14310), two for a recessive disease (e.g. Tay-Sachs Disease - 27280), and three for an X-linked trait (e.g. Lesch Nyhan Syndrome - 30800).

Verified: b if the defect was verified on the cell culture before freezing, A if verified after recovery from liquid nitrogen. Space left blank if verified on another cell culture or tissues from the same patient or relative, if not verified at all, or if verification is not yet possible because defect is unknown or not yet expressed in culture.

Paris Nomenclature: As described (Paris Conference 1971) and Supplement (1975): Standardization in Human Cytogenetics. Birth Defects: Original Article Series, VIII:7, 1972. The National Foundation, New York.

*McKusick, Victor. Mendelian Inheritance in Man 4th Edition. John's Hopkins Press, Baltimore, London. 1975.

Balanced, Unbalanced: Balanced (B) or Unbalanced (U) karyotype

Remarks: Any pertinent information not included in preceding columns.

*after GM #: Means a description of this cell culture has been published in Cytogenetics and Cell Genetics. Reference numbers appearing in parentheses at the bottom of the page refer to these publications listed in Appendix C, pages 153-160.

Aging Cell Repository: Cell cultures listed under this heading are of particular interest to investigators interested in studies on aging. Some cultures originally deposited in the GM Repository are also of interest to the Aging Repository. For convenience these cell cultures are listed in both the GM and the Aging (AG) Repository. Many new cell cultures are now being added to the Aging Repository and receive an AG #. Double listings will be made when appropriate. Requests for these cells may be addressed to the Director of the Aging Repository:

Dr. Warren W. Nichols
Institute for Medical Research
Copewood Street
Camden, New Jersey 08103
Telephone 609-966-7377

Procedures and charges are the same as for the Genetic Mutant Cell Repository. The Aging Repository is supported by the National Institute of Aging.

HUMAN FIBROBLAST CULTURES WITH
BIOCHEMICAL MUTANT CONDITIONS

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF AMINO ACID METABOLISM					
Alkaptonuria - 20350									
2228	4	B	147	36	M	W	--		
Argininemia - 20780									
954	5	C	6	4	F	W	--		
Argininosuccinicaciduria - 20790									
525	8	C	123	9	M	W	--	A	Family
533	8	C	123	20	M	W	--	A	
540	8	C	123	63	M	W	+-	B	
Citrullinuria (Citrullinemia) - 21570									
63	17	S	95	9 mo.	F	W	--	B	ATCC CCL 76
1044*	3	A	107	8 mo.	M	W	--	A	Proband; see GM-1204 Lymphoid
1058	3	A	107	27	M	W	+-(0)		Father; see GM-1206 Lymphoid
1679	2	B	157	1 da.	M	W	--		A sib died of Citrullinemia; See also GM-1685 Lymphoid
1684	6	C	114	1 da.	F	W	--	B	Neonatal
Cystinosis									
Type I - (Early Onset Nephropathic Type; Infantile Type) - 21980									
8	5	A	121	5	F	W	--	A	
18	4	A	121	3	M	W	--	A	
20	4	A	121	5 mo.	M	W	--	A	
46	5	A	121	3 1/2	M	W	--	A	
489	10	G	114	1 1/2	M	W	--		Sib of GM-304

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #46

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF AMINO ACID METABOLISM					
Cystinosis									
Type I, continued									
304	4	A	114	6 mo.F	M	W	--		Proband, skin; formerly GM-91 — Same fetus
90	3	A	114	6 mo.F	M	W	--		Lung — Same fetus
93	4	A	114	6 mo.F	M	W	--		Umbilical Cord — Same fetus
706	3	A	117	1 1/2	M	W	--		
760	2	A	128	6 mo.F	M	W	--		Fibroblasts also available from bone marrow, kidney, thymus, liver, thyroid and amnion
2066	2	B	19	1	F	W	--	A	Biopsy taken at autopsy
Type III - (Benign Type; Adult Type) - 22000									
378	8	G	114	7	F	W	--	B	Sib — Family
379	8	G	114	4	M	W	--	B	Sib — Family
906	11	B	114	32	M	W	+-(0)		Father — Family
907	15	B	114	30	F	W	+-(0)		Mother — Family
Type Unclassified									
908	8	B	114	12	M	W	--	B	Nephropathic — Family
909	9	B	114	42	F	W	+-(0)		Mother — Family
910	9	B	114	49	M	W	+-(0)		Father — Family
Homocystinuria (Cystathionine Synthase Deficiency) - 23620									
342	3	A	66	20	F	W	--		B6 Responsive
423	8	B	114		M		--	A	
424	9	B	114		M		--	A	
584	3	A	38	39	M	W	--	A	Proband — Family
585	3	A	38	65	M	W	+-(0)		Father — Family

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF AMINO ACID METABOLISM					
Homocystinuria, continued									
594	2	A	86	23	M	W	--	A	Proband
417	4	A	42	42	F	W	+-(0)		Mother
625	3	A	38	35	M	W	+-	A	
720	3	A	38	30	M	W	--		
721	7	A	38	11	F	W	--	B	
722	7	A	38	24	M	W	--	B	Proband
725	7	A	38		F	W	+-(0)		Mother
724	14	A	38		M	W	--		
751	3	A	86	19	M	W	--	A	Sib
752	3	A	86	23	M	W	--	A	Sib
976	3	C	86	6	F	W	+?	A	Sib
753	3	C	86	41	M	W	+-	A	Father
813	2	C	38	13	M	W	+-	A	
864	12	A	38	13	F		--	A	
865	5	A	38		M		--	A	
866	8	A	38	10	F		--	A	Sib
867	10	C	38	6	M		--	A	Sib
885	3	C	86	17	M	W	--	A	Proband
883	2	C	38	42	F	W	+-(0)		Mother
1128	3	C	38	11	F	W	--	A	Proband; B6 non-responsive
1129	3	C	38	9	M	W	--		Sib
1126	3	C	38		M	W	+-(0)		Father
1374	4	A	38	13	F	W	--	A	Proband
1375	6	A	38		F	W	+-(0)		Mother
1376	6	A	38		M	W	+-(0)		Father

Family

Family

Family

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF AMINO ACID METABOLISM					
Hyperglycinemia, Non-Ketotic - 23830									
747	3	A	107	1 1/2	F	W	--		
880	5	C	95	21	M	W	--		Sib
1140	3	A	95	16	M	W	--		Sib Family
1162	3	A	95	28	M	W	--		Sib
1297	8	B	114	6	M		--		
1298	8	B	114	4 1/2	M		--		
1299	7	B	114	2	M		--		
1300	8	B	114	2 1/2	F		--		
1301	8	G	114	2 mo.	F	W	--		
1302	9	B	114	1	F		--		
Hypermethioninemia - 23890									
911	2	C	86	3 mo.	M	W	--		
Hyperphenylalaninemia									
6	7	A	95	5	F	W	--		Proband
4	9	A	95	33	M	W	+-(O)		Father Family
7	7	A	95	30	F	W	+-(O)		Mother
Isovalericacidemia - 24350									
427	7	J	13	9 da.	M	B	--		
947	5	C	2	13	M	W	--	B	
Maple Syrup Urine Disease - (Branched-Chain Ketoaciduria) - 24860									
296	15	A	33	5	M	B	--	B	
297	18	A	33	13 da.	F	W	--	B	
16	4	A	95	25	M	W	+-(O)	A	
22	5	A	95	25	F	W	+-(O)	A	
612	2	A	74	2 mo.	F	W	--	A	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF AMINO ACID METABOLISM					
Maple Syrup Urine Disease, continued									
649	3	A	38	9 mo.	M	W	--		Proband
650	3	A	38		M	W	+-(O)		Father — Family
651	3	A	38		F	W	+-(O)		Mother
1000	10	A	27	13	F	W	--	B	Iraqi
1099	3	A	107	7 mo.	F	W	--		
1158	2 IMR	A	86	6	F	W	--	A	Proband, mild variant
1159	2 IMR	A	86		M	W	+-(O)		Father
1364	2	E	107	7	F	B	--		See GM-1366 Lymphoid
1557	12	G	27	5	M	W	--		Variant
1654	2	A	107	5	F	W	--	A	See GM-1655 Lymphoid; two sibs died of MSUD
1744	11	B	77	13 da.	F	W	--	A	
1938	7	B	77	5	M	W	--	A	
2327	3	B	107	4 mo.	F	W	--	A	Pericardium
Methylmalonicaciduria - 25100									
50	10	S	95	1	M	W	--	A	Unresponsive to B12; ATCC CCL 124
212	6	A	95	1	M	W	--	A	Responsive to B12
306	6	A	95	2	M		--	A	Responsive to B12
595	2	A	88	7	M	W	--		Proband; Cobalamin A mutant
596	2	A	88	25	F	W	+-(O)		Mother
930	6	B	32	2	F	W	--		Mutase defect; unresponsive to B12
876	5	I	10	1 mo.	M	W	--		Cobalamin B mutant
1673	8	J	88	1 da.	M	W	--	B	Apomutase mutant
1674	7	C	88	1 da.	F	W	--		Cobalamin A mutant
Phenylketonuria - 26160									
937	2	C	107	19 mo.	M	W	--		
2406	2	B	74	2 mo.	M	W	--		Pyloric stenosis

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF AMINO ACID METABOLISM					
Propionicacidemia - (Hyperglycinemia with Ketoacidosis & Leukopenia) - 23200									
56	10	A	88	1	F	W	--	B	
57	10	A	88	6	F	W	--	B	
371	3	A	105	2 mo.	F	W	--	B	Proband
403	7	A	105	24	F	W	+-(0)		Mother — Family
405	7	A	105	25	M	W	+-(0)		Father
Tyrosinemia - 27670									
286	7	A	95	2 mo.	M		--		
				DISORDERS OF CARBOHYDRATE METABOLISM					
Amyloidosis - Dominant									
1998	3	B	155	16	F	W	+-(0)		Non-Portuguese
Aspartylglycosaminuria - 20840									
568	2	A	4	18	F	W	--		
2056	3	B	4	32	M	W	--	B	
2057	4	B	4	26	F	W	--	B	
Fructose -1,6-Diphosphatase Deficiency - 22970									
282	6	A	95	7	F		--		
Fucosidosis - 23000									
289*	3	A	78	31	M	W	+-(0)		Father — Family;
290*	3	A	78	29	F	W	+-(0)		Mother — normal karyotypes
291*	3	A	78	9	M	W	--	A	Son — Fucosidosis Type II
292*	3	A	78	4	M	W	--		Son — See GM-1023, GM-1024, GM-1025, GM-1026, Lymphoid
801	6	C	138	15	M	W	--	B	Type II; Spanish-American

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #24

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF CARBOHYDRATE METABOLISM					
Fucosidosis, continued									
802	5	C	138	7 1/2	M	W	--	B	Type II; Spanish-American
1214	5 IMR	A	108	18	M	W	--		Type II
Galactosemia (Kinase Deficiency) - 23020									
334	5	A	95	22	F	W	--	A	Proband]
335	3	A	95	53	F	W	+-(0)		Mother] Family
336	4	A	95	62	M	W	+-(0)		Father]
Galactosemia (Transferase Deficiency) - 23040									
52*	11	S	95	8	M	B	--	B	Sib; G6PD Type B; ATCC CCL 133; see also GM-639, SV40 Trans.
1908	2	A	95	15	M	B	--	B	Same patient as GM-52
53*	12	S	95	6	M	B	--	B	Sib; G6PD Type A; ATCC CCL 132; see also GM-638, SV40 Trans.
54	10	A	95	3 mo.	M	W	--	B	Same]
1907	3	A	95	6	M	W	--	B	patient]
264	11	J	95	2 1/2	M	W	--	B	Duarte variant
422	3	A	95	9	F	W	--		
433	3	A	95	10	F	B	--		
438*	6	A	69	46	M	W	+-(0)	B	Father]
439*	6	A	69	41	F	W	+-(0)	B	Mother
440*	6	A	69	16	F	W	--	B	Daughter] Family
441*	6	A	69	15	M	W	--	B	Son
442*	6	A	69	12	M	W	++	B	Son
1533	7	A	69	19	F	W	++	B	Daughter]
528	4	A	95	2	F	W	--	B	
727	3	A	95	2 mo.	F	W	--	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 25; 34

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF CARBOHYDRATE METABOLISM					
Galactosemia (Transferase Deficiency), continued									
1208	3	C	45	27	M	W	+-	A	Father
1209	2	C	45	1	M	W	--	A	Twin; Proband
1210	2	C	45	1	M	W	--	A	Twin; Proband
1211	3	C	45	4	M	W	+?	A	Son
1212	3	C	45	29	F	W	+-	A	Mother
1417	6	A	95	22	F		--		Proband
1418	5	A	95		F		+-(O)		Mother
1419	5	A	95		M		+-(O)		Father
1703	4	B	95	9 mo.	M	W	--	B	Cousin
1704	3	B	95	2	M	W	--	B	Cousin
1741	8	B	77	6 mo.	M	W	--	B	See GM-1743, amniotic fluid cell
1996*	6	J	2	1	M	W	--	A	Double heterozygote; GALT, Duarte
Glucose-6-Phosphate Dehydrogenase Deficiency and Variants (See Biochemical Markers) - 30590									
120	9	C	41	30	F	B			G6PD A/B; PGK 1,2
218	9	C	41	12	M	W	Gd-		Ca. zero activity
324	5	A	125	22	M	W	Gd-/Gd-		47,XXY; Mediterranean type
325	4	A	125	30	M	W	Gd+/Gd-		47,XXY; Mediterranean type
412	3	A	9	19	M	W	Gd-		Panama type
738	7	A	41		M	W	Gd+		Hektoen variant
888	22	C	125	2 1/2	M		Gd-		New York type
Glycogen Storage Diseases									
Type I - Von Gierke Disease - 23220									
574	8	C	130	25	M	W	--		

Family

Family; slow growing

2nd generation with GALT

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #48

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF CARBOHYDRATE METABOLISM					
Glycogen Storage Diseases									
Type II - Pompe Disease - 23230									
244	4	A	2	5 mo.	F	W	--	A	Mexican
248	3	A	95	4 mo.	M	B	--	A	
338	2	A	8	18 da.	M	B	--		Grows poorly
443	4 IMR	A	99	30	M		--	A	Late onset
1935	3	A	126	30	F	B	--	A	
Type III - Debranching Enzyme Deficiency - 23240									
111	7	A	143	1	F	W	--	A	
226	12	A	143	13	F		--	A	
303	9	A	95	9	F	W	--	A	
573	7	C	62	16	F	W	--	B	
576	6	C	62	7	F	O	--	B	
578	18	C	62	4	M	W	--	B	
683	3 IMR	C	95	9 mo.	M		--		
1702	4	B	95	11	M		--	A	
Type IV - Andersen Disease - 23250									
572	9	C	62	2	F	W	--	A	
Type V - McArdle Disease - 23260									
577	7	C	130	39	M	W	--		
Type VI - Hers Disease - 23270									
579	9	C	62		M		--		
Types VIII, IX - Phosphorylase Kinase Deficiency - 30600									
25	4	A	95	2 1/2	M	W	y-		
575	5	C	62	3	M	W	y-		Mexican

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF CARBOHYDRATE METABOLISM					
Hyperpyruvicacidemia									
583	3	A	38	7 mo.	F	W	--		
Lactic-pyruvic Acidosis									
2224	2	B	162	1 wk.	F	W			
Mannosidosis - 24850									
654	3	A	4	7	M	W	--	A	
1851	10	A	6	3	M	W	--	B	
2049	8	G	77	39	M	W	+-	B	Father]
2050	9	B	77	11	M	W	--	B	Proband Family - New variant
2051	18	B	77	7	F	W	--	B	Sister]
Mucolipidoses									
Type II - I-Cell Disease - 25250									I-Cell disease lines are difficult to recover from liquid nitrogen
87	5	A	77	5 mo.	M	W	--		Proband]
521	4	A	95	2	M	W	--	A	Same patient as GM-87
80	5	A	95	17	F	W	+-(0)		Mother Family
81	5	A	95	23	M	W	+-(0)		Father]
164	4	A	105	5 1/2	M	W	--	A	
1586	9	B	77	1 mo.	M	W	--		Proband]
1589	7	B	77		M	W	+-(0)		Father Family
1590	7	B	77		F	W	+-(0)		Mother]
1494	6	A	57	16	F	W	--	A	Atypical mild variant
1742	3	B	77	4 mo.	F	W	--	B	
2013	8	U	77	1 mo.	M	W	--	B	Proband]
2014	8	B	77	18	F	W	+-	B	Mother]

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF CARBOHYDRATE METABOLISM					
Mucolipidoses									
Type II - I-Cell Disease, continued									
2045	7	H	95	1 da.	M	W	--	B	Proband
2046	6	B	95	25	F	W	+-	B	Mother — Family
2047	4	A	95	26	M	W	+-	B	Father
2145	8	B	77	26	M	W	+-(0)		Father of I-cell
2273	3	B	70	2	M	W	--		Proband
2274	3	B	70		F	W	+-(0)		Mother
Type III - Pseudo-Hurler Polydystrophy - 25260									
2065	3	B	126	9	M	W	--	B	
2425	6	C	134	15	M	W	--	B	
113	4	A	105	2 1/2	F	W	--	A	
1759	2	B	92	13	M	W	--	B	
Type IV									
2048	10	B	77	2	F	W	--	B	
Mucopolysaccharidoses									
Type IH - Hurler Syndrome - 25280									
2	7	A	95	10 mo.	M	W	--	A	Proband
3	8	A	95	26	F	W	+-(0)		Mother
34	6	A	95	9 mo.	F	W	--	A	Proband
31	4	A	95	23	F	W	+-(0)		Mother — Family
42	5	A	95	25	M	W	+-(0)		Father

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF CARBOHYDRATE METABOLISM					
Mucopolysaccharidoses									
Type IH - Hurler Syndrome, continued									
415	2	A	107	4	M	W	--	A	
798	3 IMR	C	62	1	F	W	--	A	Proband
799	3 IMR	C	62	22	F	W	+-(0)		Mother — Family
800	3 IMR	C	62	27	M	W	+-(0)		Father
887	5	C	57	7	F	W	--	B	
1053	5	C	57	4	M	W	--	B	
1257	7	K	62	1	F	W	--	A	
1391	2	A	70	9 mo.	F	W	--	A	Proband
1392	3	A	70	24	F	W	+-(0)		Mother — Family
1393	3	A	70	24	M	W	+-(0)		Father
Type IH/S - Hurler/Scheie									
512	4	A	8	15	F	O	--		
963	3	C	92	5 1/2	M	W	--	A	
1254	9	C	62	4	M	W	--	A	Sib
1255	6	K	62	10	M	W	--	A	Sib
1898	10	B	77	6	M	W	--	B	Proband
2016	6	B	77	35	F	W	+-	B	Mother — Family
2017	6	B	77	40	M	W	+-	B	Father
Type IS - Scheie Syndrome - 25310									
1256	9	K	62	12	M	W	--	A	
1323	3	A	92	58	M	W	--	A	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF CARBOHYDRATE METABOLISM					
Mucopolysaccharidoses									
Type II - Hunter Syndrome - 30990									
39	4	A	66	9	M	W	y-	A	
47	6	A	107	9	M	W	y-	A	
140	4	C	105	5	M	B	y-	A	
298	4	A	66	2	M	W	y-	A	
614	3	A	92	18	M	W	y-	A	Proband; mild type
613	3	A	92		F	W	+-(0)		Mother
615	3	A	92	9	M	W	y-	A	Proband; severe type
620	3	A	92		F	W	+-(0)		Mother
690	3	A	92	14	M	W	y-	A	
862	2	C	107	11	M	W	y-	A	Mild type
901	11	C	57	5	M	W	y-	B	Proband
902	6	C	57	28	F	W	+-(0)		Mother
1258	7	K	62	2	M	W	y-	A	
1583	7	B	77	3 1/2	M	W	y-	A	
1927	5	C	96	16	M	W	y-	B	Mild type; uncle
1928	5	C	96	7	M	W	y-	B	Mild type; nephew
1929	5	C	96	7	M	W	y-	B	Severe type
2268	8	A	168	5	F	W	-?	A	Rare in females
Type IIIA - Sanfilippo Syndrome, A - 25290									
312	2	A	107	3	F	W	--	A	
629	3	A	66	10 1/2	M		--	A	
643	3	A	107	3 1/2	M	W	--	A	
879	7	C	57	3	F	W	--	A	Proband
886	7	C	57	19	F	W	+-(0)		Mother
903	8	C	57	10	M	W	--	A	
934	2 IMR	C	70	7	F	W	--	A	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks	
				DISORDERS OF CARBOHYDRATE METABOLISM						
Mucopolysaccharidoses										
Type IIIA - Sanfilippo Syndrome, A, continued										
1094	3	A	95	5 1/2	F	W	--	A	Proband	
1739	3	A	95	6 1/2	F	W	--		Same patient as GM-1094	
1095	4	A	95		F	W	+-(O)		Mother	Family
1096	3	A	95		M	W	+-(O)		Father	
Type IIIB - Sanfilippo Syndrome, B -25292										
156	5	A	107	7	M	W	--	A		
737	3	A	107	7	M	W	--	A		
1426	3	A	70		F		--			
Type IV - Morquio Syndrome - 25300										
593	3	A	92	7 1/2	F	W	--	A		
958	2	C	92	12	M	W	--	A	Clinically atypical	
1259	9	K	62	14	F	W	--		Clinically atypical	
1361	3	A	92	43	M	W	--	A	Variant	
1602	3	A	66	11	F	W	--	B		
Type VI - Maroteaux-Lamy Syndrome - 25320										
519*	9	A	8	4	F	W	--	A	Proband; see GM-1022 Lymphoid	
520*	8	A	8	42	M	W	+-	A	Father	
935*	6	C	8	40	F	W	+-	A	Mother	Family
943*	7	C	8	8	F	W	+-	A	Sib	
942*	7	C	8	5	F	W	++	A	Sib	
538	9	A	105	1 1/2	F	W	--	B		
552	11	A	105	14	M	W	--			

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #35

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF CARBOHYDRATE METABOLISM					
Mucopolysaccharidoses									
Type VII - Beta-Glucuronidase Deficiency - 25322									
151	3	A	66	4 1/2	M	W	--	A	
121*	3	A	126	3	M	B	--	A	Proband
1850	3	B	126	28	M	B	+-	A	Father — Family
2074	4	B	126	21	F	B	+-(O)		Mother
Winchester Disease - 27795									
2295	2	B	180	21	F	P	--		
Neuraminidase Deficiency									
1718	2	B	70	2 mo.	F		--		Proband
1719	3	B	70	25	F		+-(O)		Mother — Family
1720	2	B	70	26	M		+-(O)		Father
Phosphoglycerate Kinase Deficiency - 31180									
743	2	A	98	4	M	O	y-		
				DISORDERS OF LIPID METABOLISM					
Carnitine Palmitoyl - CoA Transferase Deficiency									
1763	4	A	95	29	M	W	--		Formerly GM-249
Ceroid-lipofuscinosis - 16235									
741	6	C	66	5	M		--		
Cholesterol Ester Storage Disease of Liver - 21500									
863	5	C	6	11	F	W	--	B	
Fabry Disease (Diffuse Angiokeratoma) - 30150									
107	5	A	105	10	M		y-	A	
881	9	C	108	17	M	W	y-	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #32

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF LIPID METABOLISM					
Fabry Disease, continued									
882	10	C	108	40	M	W	y-	B	
1068	9	A	32	35	M	W	y-	B	Proband]
1070	8	A	32	52	F	W	+-	B	Mother] Family
Farber Lipogranulomatosis - 22800									
904	14	C	105	8 mo.	F	W	--		Proband]
994	11	A	105	29	F	W	+-(0)		Mother] First] Family
995	11	A	105	36	M	W	+-(0)		Father] cousins]
2315	8	A	175	7	F	W	--		Italian descent
2314	4	A	175	6	F	W	--		Proband; Irish descent]
2316	2 IMR	A	175	37	M	W	+-(0)		Father] Family
2317	2 IMR	A	175	37	F	W	+-(0)		Mother]
Gaucher Disease									
Type 2 (Infantile, Cerebral) - 23090									
855	10	C	138	4 mo.	F	W	--	B	
877	2	A	107	1	M	W	--	A	Proband]
878	2	C	107		F	W	+-(0)		Mother]
1260	2	A	50	11 mo.	F	W	--	A	
Types 1 & 3 (Juvenile & Adult) - 23100									
372	4	A	9	29	M	W	--		
852	5	C	138	20	M	W	--	B	
1607	3	A	47	30	M	W	--	A	
GM1 Gangliosidosis - Type I - 23050									
806	3	A	126	2 1/2	F	W	--	A	
918	18	C	49	1	M		--		From Canadian Repository

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF LIPID METABOLISM					
GM2 Gangliosidoses									
AB Variant									
1675	5	C	8	16 mo.	F	B	--	B	
Type I - Tay-Sachs Disease - 27280, 27290									
77	9	A	95	1	M	B	--	A	Non-Jewish
221	4	A	66	3	M	W	--	A	
502	2	A	66	11 mo.	M	W	--		
514	8	C	68	3	F	W	--	B	Sib; Pericardium
515	5	C	68	1	F	W	--	A	Sib
527	3	A	107	1 1/4	M	W	--		
1108	8	A	68	30	F	W	+-(0)		Mother
1109	7	C	68	31	M	W	+-(0)		Father Family;
1110	7	C	68	10 mo.	M	W	--	B	Proband Non-Jewish
Type II - Sandhoff Disease - 26880									
203	5	A	107	1	M	W	--	A	Proband
204	5	A	107	18	F	W	+-(0)		Mother
294	5	I	17	2	M	W	--		
317	3	A	66	1	F	B	--	A	
470	8	C	9	1	F	W	--		
2094	2	B	181	12	M	W	--	B	Juvenile; variant
Hyperlipoproteinemia									For types other than II, inquire
Type II - Familial Hypercholesterolemia - 14440									
283	5	A	45	28	F		+-	A	Mother
376	5	A	45	6	F		+-	A	Daughter

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF LIPID METABOLISM					
Hyperlipoproteinemia									
Type II - Familial Hypercholesterolemia, continued									
488	4	A	84	11	M	W	--	A	Proband; receptor negative
483	4	A	84	37	F	W	+-	A	Mother
966*	11	S	50	12	F	W	--	A	Receptor negative; formerly GM-361*
486	4	A	84	25	F	W	--	A	Receptor negative
1116	9	J	50	7	F	W	--	A	Receptor negative Same
2000	2	B	50	7	F	W	--	A	Receptor negative patient
701	7	B	72	6	F	W	--	A	Proband; receptor negative
700	7	B	72	45	F	W	+-(0)		Mother; other family members are available upon inquiry from Repository
1915	3	B	50	13	F	B	--	A	Receptor negative
Krabbe Disease (Globoid Cell Sclerosis) - 24520									
267	7	I	8	36	F	W	+-(0)		
268	6	I	58	41	M	W	+-(0)		
853	8	C	138	1	F	W	--		
854	8	C	138	9 mo.	F	W	--		
1773	2	B	105	1	M	W	--	A	
Metachromatic Leukodystrophy									
Infantile - 25010									
78	7	A	131	3	M	W	--	A	
243	2	A	74	3 1/2	M	W	--	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #26

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks	
				DISORDERS OF LIPID METABOLISM						
Metachromatic Leukodystrophy										
Infantile, continued										
197	3	A	105	4	M	W	--	A	Proband	Family
196	3	A	105	33	M	W	+-(O)		Father	
754	8	A	105	37	F	W	+-(O)		Mother; grows poorly	
200	4	A	105	5	M	W	+-	B	Brother	
560	5 IMR	A	105	27	F	W	+-	B	Aunt	
357	9	A	105	3	M	W	+-	B	First cousin	
561	5 IMR	A	105	54	F	W	+-	B	Grandmother	
640	10	A	105	25	F	W	+-	B	Aunt; Mother of GM-357	
905	2	C	122	3	F	W	--	A		
2093	3	B	105	25	M	W	--			
2331	5	A	134	5 1/2	M	W	--	A		
Adult - 25000										
132	6	A	105	22	M	W	+-	B	Sib	
133	5	A	105	19	M	W	+-	B	Sib	
Multiple Sulfatase Deficiency Disease										
915	9	C	105	2	M	W	--	B		
2407	10	A	147	3	F	W	--	B		
Niemann-Pick Disease										
Type A - 25720										
112	3	A	95	10 mo.	M	W	--	A		
370	8	K	2	1	F	W	--	A		
406	9	A	105	2	F	W	--	A		
559	4 IMR	A	105	25	F	W	--	B		
641	19	A	105	4	M	W	--			
644	5	A	105	3	F	W	--	B		
898	3	A	105			W	--			

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF LIPID METABOLISM					
Niemann-Pick Disease									
Type B - 25720									
1669	12	C	105		F	W	+-(O)	B	
Type C - 25725									
110	6	A	107	9	M	W	--		
645	8	C	105	4	F	W	--	B	
Type Unspecified									
165	3	A	107	7	M	W			Clinically atypical
1612	8	C	138		M	W	--	B	Variant; sib
1613	7	C	138		M	W	--	B	Variant; sib
Refsum Syndrome - 26650									
1007	17	C	129	30	F	W	--	B	
Schilder Disease - 26910									
269	4	A	58	30	F	W	+-(O)		
Wolman Disease - 27800									
1606	4	B	154	7 mo.	F	W	--	B	
2211	4	H	187	21 wk.F	M	W	--		Proband
2121	7	A	187	31	F	W	+-(O)		Mother
				DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM					
Adenosine Deaminase Deficiency (Immune Deficiency Disease) - 24275									
469	8	A	118	1	M	W	--	B	
471	4	A	103	1 1/2	F	B	--	B	
2027	9	A	118		F		+-	B	
2028	11	A	118		M		+-	B	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM					
Adenosine Phosphoribosyltransferase Deficiency - 10260									
517	5	C	71	64	M	B	+-		Enzyme deficient in erythrocytes; normal level in fibroblasts
Inosine Triphosphate Pyrophosphohydrolase (ITPase) Deficiency									
1617	2	B	177	29	F	W			See GM-1619 Lymphoid
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800									
377	8	A	77	8 mo.	M	W	y-	B	Proband] Family
13	7	A	95	34	F	W	+-	B	Mother]
1906	3	A	95	15	M	W	y-		Proband]
14	6	A	95	37	F	W	+-(0)		Mother]
68	14	J	27	2	M		y-	A	
152	10	B	101	9	M	W	y-	A	
158	9	B	101	3	M	B	y-	A	Proband; G6PD Type A]]
135	8	A	101		F	B	+-	A	Mother; G6PD Type AB; HGPRT + coupled with G6PD B; HGPRT - coupled with G6PD A; sister of GM-318]
									Family]
159	11	B	85	10	M	B	y-	A	Sib; G6PD Type A
2063	10	J	101	5 1/2	M	B	y-	A	Sib; G6PD Type A; replaces GM-177; see also GM-847, SV40 Trans.
318	13	C	101	40	F	B	+-	A	Mother;G6PD Type AB; HGPRT + coupled with G6PD B; sister of GM-135]]

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM					
Lesch-Nyhan Syndrome (HGPRT Deficiency), continued									
1390	5	B	90	3 mo.	M	W	y-	A	
537	7	B	31	13	M		y-	A	
1362	3	A	66	10	M	W	y-	A	
1662	2	B	66	9	M	W	y-	A	Variant - Proband
1659	3	B	66	42	F	W	+-	B	Mother
1661	3	B	66	14	F	W	+-	B	Sibling Family
1660	3	B	66	11	F	W	+-	B	Sibling
2226	3	B	66	40	F		+-(O)		Mother
2227	3	B	66	12	M		y-		Proband
2290	2	B	183		M		y-		Skin same fetus; see also
2291	2	B	183		M		y-		Lung GM-2292 Lymphoid & GM-2338 Amniotic
Orotic Aciduria - 25890, 25892									
328	22	A	125	2	M	W	--	B	Medium should contain uridine
632	2	C	115	14	F	W	--		Medium should contain uridine
Xeroderma Pigmentosum - 27870									
Complementation Group A									
518	19	C	23	1	M	W	--	B	De Sanctis-Cacchione XP25RO* ATCC CRL 1261
544	2	A	23	10	M	W	--		De Sanctis-Cacchione XP4LO
710	11	A	11	1 1/2	M	W	--	B	De Sanctis-Cacchione XP26RO
Complementation Group C									
30	3	A	110	24	M	W	--		XP3BE; ATCC CRL 1189

*Nomenclature as per 13th International Congress of Genetics 79:215-225, 1975

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM					
Xeroderma Pigmentosum									
Complementation Group C, continued									
671	4	A	23	15	M	W	--		XP8BE; ATCC CRL 1158; twin
676	3	A	23	15	M	W	--		XP9BE; ATCC CRL 1161; twin
673	4	A	23	21	M	W	--		XP4SL; sib
677	3	A	23	25	M	W	--		XP2BE; ATCC CRL 1166; sib
709	17	A	11	15	M	W	--	B	XP21RO
Complementation Group D									
434	3	A	23	28	M	W	--		De Sanctis-Cacchione XP3NE
435	3	A	23	23	F	W	--		De Sanctis-Cacchione XP2NE
Complementation Group E									
1762	15	A	11	34	F	W	--	B	XP2RO; formerly GM-708; ATCC CRL 1259
Complementation Group Undetermined									
82	5	A	44	12	F	W	--		Proband
241	11	A	44		F	W	+-(O)		Mother
436**	3	A	23	22	F	W	--		XP1NE
510	9	J	23	10	M	W	--	A	XP1PW
522**	6	C	37	16	F	B	--		G6PD Type B; phosphoglucomutase I; XP2Nbi
523**	9	A	37	10	F	B	--	A	G6PD Type B, phosphoglucomutase I; XP1Nbi
936**	8	C	46	22 wk.F	F	W	--		Umbilical cord
1213	2	C	133	61	F	W	--		
1227	3	A	133	28	M	W	--	A	Variant, high repair activity
1295	4	C	145	6	F	W	--	A	XP1KC
1389	4	A	133	21	F	W	--	A	See GM-1646 Lymphoid

**Low in unscheduled DNA synthesis

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM									
Xeroderma Pigmentosum									
Complementation Group Undetermined, continued									
1509	4	A	133	38	F	W	--		
1630	2	A	173	3 1/2	M	W	--	A	Proband]
1631	2	A	173	29	M	W	+-	A	Father] Family
1632	3	C	173	25	F	W	+-	A	Mother]
2024	2	B	170	3	M	B	--		Sib]
2034	4	B	170	28	F	B	+-(0)		Mother] Family
2035	3	B	170	5	M	B	--		Proband]
OTHER DISORDERS OF KNOWN BIOCHEMISTRY									
Acatalasia (Acatalasemia) - 20020									
64	5 IMR	L	80		M	W	--	B	Swiss]
65	7 IMR	L	80		M	O	--	B	Japanese] Limited supply
66	7 IMR	L	80	60	M	O	--	B	Japanese]
1931	8	A	169	73	M	W	+-(0)		
Menkes Syndrome (Kinky Hair Disease) - 30940									
220	4	A	74	2 mo.	M	W	y-	A	Sib]
057	3	A	107	5 da.	M	W	y-		Sib; see GM-1245 Lymphoid] Family
245	3	A	74	1 mo.	M	W	y-	B	Cousin]
1981	3	B	107	2	M	W	y-	B	Proband; see GM-1982 Lymphoid]
1983	3	B	107	26	F	W	+-	B	Mother; see GM-1984 Lymphoid]
Porphyria									
Acute Intermittent Porphyria - 17600									
931	3	B	112	32	F	W	+-	A	Mother]
932	3	B	112	4	M	W	+-	A	Son] Family
933	3	B	112	1 1/4	F	W	+-	A	Daughter]

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				OTHER DISORDERS OF KNOWN BIOCHEMISTRY					
Porphyria									
Acute Intermittent Porphyria, continued									
939	3	C	112	32	F	W	+-	A	Daughter; see GM-2135 Lymphoid
940	2	C	112	61	F	W	++	A	Mother; see GM-2134 Lymphoid
941	3	C	112	63	M	W	+-	A	Father; see GM-2133 Lymphoid
1621	2	B	112	39	F	W	+-	A	Mother
1622	3	B	112	19	F	W	+-	A	Sister Family
1623	2	B	112	16	M	W	+-	A	Son
1624	3	B	112	22	F	W	+-	A	Mother
1625	3	B	112	3	M	W	+-	A	Son
1647	3	B	112	44	F	W	+-	A	
Cutanea Tarda - 17610									
961	3	A	112	70	M	W	+-		
977	5	B	112	81	F	W	+-		
1041	6	B	112	69	F	W	+-		
1082	2	B	112	21	M	W	+-		
1179	3	B	112	36	M	W	+-		
1482	4	B	112	58	M	W	+-		
Hereditary Coproporphyria - 12130									
962	3	C	112	59	F	W	+-		
Sickle Cell Anemia (Hemoglobin S) - 14170									
2340	10	J	107	4 mo.	F		+-		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY					
Adrenal Hyperplasia (Adrenogenital Syndrome) Type III - 20191									
2241	2	B	107	9 mo.	F	W	--		See GM-2242 Lymphoid
Adrenoleukodystrophy (Addison Disease and Cerebral Sclerosis) - 30010									
623	8	A	105	12	M	W	y-		Proband
337	3	A	105	35	F	W	+-(0)		Mother
Agammaglobulinemia - 30030									
362	8	A	125	21	M	W	y-		
Alzheimer Disease of Brain - 10430									
364	2	A	139	53	M	W	+-(0)		Aunt; see GM-364 Aging Repository
490	3	A	139	33	F	W	+?		Niece
Basal Cell Nevus Syndrome - 10940									
1552	2	B	104	27	M	W	+-(0)		
1577	6	B	104	39	M	W	+-(0)		
1658	3	B	104	53	M	W	+-	B	Basal cell carcinoma; see GM-1656 Lymphoid
1657	3	B	104	53	M	W	+-	B	Normal skin (Same patient)
1725	3	B	104	58	F	W	+-(0)		See GM-1726 Lymphoid
2098	3	B	104	31	M	W	+-(0)		See GM-2099 Lymphoid
2138	4	B	104		F	W	+-(0)		See GM-2139 Lymphoid
Beckwith-Wiedemann Syndrome (EMG Syndrome) - 22560									
359	3	A	107	8	M	W	--		
Campomelic Dwarf - 21135									
1737	9	A	142	9 da.	M	W	--		Karyotypic abnormalities

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY					
Canavan Disease (Spongy Degeneration of the CNS) - 27190									
59	2	A	95	1	F		--		
60	3	A	95	2	M		--		
Cartilage - Hair Hypoplasia (Metaphyseal Chondrodysplasia, McKusick Type) - 25025									
1671	4	B	70	17	M		--		
CNS Disorder, Unclassified, Hereditary									
1358	3	A	70	15	M	W	--		6 of 16 siblings affected with unclassified leukodystrophy
Chediak - Higashi Syndrome - 21450									
2075	3	B	126	1 1/2	F	W	--		Clinically documented
Chondrodystrophy									
229	3	A	107	9 mo.	M	W			Mother also affected
Cockayne Syndrome - 21640									
739	3	A	115	3	F	W	--		
1098	3	C	6	20	M	W	--		
1428	9	A	90	10	F	B	--		
1629	3	B	155	10	F	W	--	B	
1856	4	B	155	13	M	W	--	B	
Conradi Syndrome (Chondrodysplasia Punctata) - 21510									
740	2	A	107	1	F	W	--		
Corneal Dystrophy, Macular Type - 21780									
1125	3	C	92	41	F	W	--		Corneal button
Cornelia de Lange Syndrome - 21790									
45	5	A	66	13	M	W	--		

DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
Cutis Laxa - 21910									
480	4 IMR	A	66	45	F	W	--		
1353	3	A	147	12 da.	M	W	--		Carrier of X-autosome translocation
1377	4	C	147	8 wk.	M	B	--		
Cystic Fibrosis (Mucoviscidosis) - 21970									
142	3	A	107	14	M	W	--		
668	11	B	59	10	M	W	--		
768	3	A	60	13	M	W	--		
770	2	A	60	19	M	W	--		
851	7	I	25	22	M	W	--		
997	8	B	59	10	M	W	--		
998	8	B	59	4	M	W	--		
999	14	B	59	13	F	W	--		
1707	10	B	5	9	F	W	--		Formerly GM-1008
1009	9	B	5	34	M	W	+-(0)		Father
1011	9	B	5	7	M	W	--		Son — Family
1012	8	B	5	8	M	W	--		Son
1708	12	B	5	33	M	W	+-(0)		Father; formerly GM-1010
1013	9	B	5	11	M	W	--		Son — Family
1014	8	G	5	13	M	W	--		Son
1348	3	B	144	18	F	W	--		
1957	5	A	178	11	M	W	--		Sib
1959	4	A	178	10	M	W	--		Sib
1958	4	A	178	13	M	W	--		
Diabetes Mellitus - 22210									Mode of transmission uncertain
609	3	A	100	15	F	W			
1828	4	C	178	18	M	W			Lipoatrophic diabetes

DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
Diabetes Mellitus, continued									
1486	3	A	34	37	M	W			Maturity onset diabetes; see GM-1241 Lymphoid
Family #1 - Maturity Onset Diabetes									
1122	3	C	34	33	F	W			Sib; see GM-1240 Lymphoid
1219	5	A	34	46	M	W			Sib; see GM-1246 Lymphoid
1237	3	C	34	29	M	W			Sib; see GM-1247 Lymphoid
1430	3	A	34	35	M	W			Sib; see GM-1243 Lymphoid
1955	3	A	34	44	M	W			Sib; see GM-1956 Lymphoid
1497	2	A	34		F	W			Sib; see GM-1498 Lymphoid
1435	5	A	34	22	M	W			See GM-1242 Lymphoid] Sons of
1496	3	A	34	20	M	W			See GM-1244 Lymphoid] GM-1497
1409	3	A	34	15	F	W			Maternal cousin of GM-1435 & GM-1496; only Juvenile Diabetic in family; see also GM-1410 Lymphoid
1837	4	B	34	22	F	W			A paternal second cousin of the 6 sibs; MODY type Group II; see also GM-1838 Lymphoid
Family #2 - Juvenile Onset with Optic Atrophy - 22230									
1609	7	A	95	18	F	W			Sib; optic atrophy; see GM-1799 Lymphoid
1610	3	A	95	13	F	W			Sib; optic atrophy; see GM-1795 Lymphoid
1611	4	A	95	15	F	W			Sib; optic atrophy
1701	4	B	95	42	M	W			Normal father; see GM-1797 Lymphoid

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY					
Diabetes Mellitus, continued									
Family #3									
1872	3	A	95	12	F	W			Affected sister
1873	3	A	95	15	F	W			Affected sister
1874	4	A	95	22	F	W			Non-diabetic sister; retarded
1875	4	A	95	17	M	W			Normal brother
1876	4	A	95	18	M	W			Normal brother
1911	3	A	95	20	M	W			Affected brother
1878	4	A	95	39	F	W			Mother
1909	3	A	95	19	M	W			Affected brother; hypertension, short stature
1910	3	A	95	21	F	W			Normal sister
Dysautonomia (Riley-Day Syndrome) - 22390									
732	2	A	107	1	M	W	--		Non-Jewish
850	5	I	25	26	M	W	--		
2341	7	C	191	17	M	W	--		Sib; mild familial type
2342	6	C	191	19	M	W	--		Sib; mild familial type
2343	6	C	191	24	F	W	--		Mild familial type
Dyskeratosis Congenita - 30500									
1774	2	B	107	6 1/2	M	W	y-		Proband; see GM-1775 Lymphoid
1786	2	B	107	30	F	W	+-(0)		Mother — Family
1787	2	B	107	78	F	W	+-(0)		Great grandmother
Dystonia Musculorum Deformans - 12810, 22450									
2215	4	B	184	30	M		+-(0)		See GM-2217 Lymphoid
2255	3	B	184	14	F		--		Recessive form; see GM-2256 Lymphoid
2304	3	B	184	16	F		+-(0)		See GM-2305 Lymphoid
2306	2	B	184	13	M	W	+-(0)		See GM-2307 Lymphoid

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
			DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY						
Ehler-Danlos Syndrome									
Type I - 13000									
1812	3	B	171		F		+-	B	
2007	6	B	171		F		+-	B	
Type II - 13000									
1691	3	B	171	19	M	W	+-	B	Same patient
2006	3	B	171	19	M	W	+-	B	
1788	2	B	171	55	M	B	+-	B	
Type IV - 13005									
2207	21	K	147	18	F	W	+-(0)		
Type VI - (Hydroxylysine-deficient Collagen) - 22540									
1790	15	C	147	12	F	W	--		Sib
1791	15	C	147	9	F	W	--		Sib
Type Unclassified									
733	3	A	143	7	M	W			
161	3	C	95	9	F				
2293	2	B	185	14	F	W			
Giant Mitochondrial Disease - 25540									
28	4	A	95	21	F	W	--		
Gouty Arthritis									
432	3	A	120	32	M	W			
Huntington Chorea - 14310									
305	5	A	35	56	F	W	+-(0)		
1061	2	A	136	51	M	W	+-(0)		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY					
Huntington Chorea, continued									
1083	3	A	136	47	F	W	+-(O)		
1085	3	A	136	44	M	W	+-(O)		
1136	4	A	136	45	M	W	+-(O)		
1168	3	C	136		M	W	+-(O)		
1169	2	C	136	50	M	W	+-(O)		
1170	4	C	136	35	F	W	+-(O)		Sib
1171	3	C	136	37	F	W	+?		Sib
1187	3	A	136	43	M	W	+-(O)		
2077	7	B	136	25	F	W	+?		Daughter
2079	3	B	136	48	F	W	+-(O)		Mother
2147	6	B	186	55	M	W	+-(O)		Proband; see GM-2146 Lymphoid
2149	3	B	186	54	F	W	++	Family	Normal spouse, see GM-2148 Lymphoid
2151	3	B	186	26	F	W	+?		Daughter; see GM-2150 Lymphoid
2153	3	B	186	40	F	W	++		Normal widow of deceased proband; see GM-2152 Lymphoid
2155	3	B	186	20	F	W	+?		Daughter; see GM-2154 Lymphoid
2157	4	B	186	16	F	W	+?	Family	Daughter; see GM-2156 Lymphoid
2159	3	B	186	19	F	W	+?		Daughter; see GM-2158 Lymphoid
2161	3	B	186	21	M	W	+?		Son; see GM-2160 Lymphoid
2163	3	B	186	12	M	W	+?		Son; see GM-2162 Lymphoid

DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks	
Huntington Chorea, continued										
2165	2	B	186	55	M	W	+-(O)		Proband	Family
2177	6	B	186	26	M	W	+?		Son; see GM-2176 Lymphoid	
2183	3	B	186	21	F	W	+?		Daughter; see GM-2182 Lymphoid	
2169	3	B	186	52	M	W	++		Normal spouse of GM-2166 Lymphoid (affected sister of proband); see also GM-2168 Lymphoid	
2187	3	B	186	60	F	W	+?		Sister of proband; see GM-2186 Lymphoid	
2189	2	B	186	63	M	W	++		Normal spouse of GM-2187; see GM-2188 Lymphoid	
2171	2	B	186	22	F	W	+?		Daughter; see GM-2170 Lymphoid	Family
2173	3	B	186	51	F	W	+-(O)		Proband; see GM-2172 Lymphoid	
2175	2	B	186	55	M	W	++		Normal spouse; see GM-2174 Lymphoid	
Hypophosphatasia - 14630										
1571	3	A	95		M	B	+-		Fetal	
Ichthyosis Congenita - 24230										
222	4	A	116	2 da.	F	W	--			
Incontinentia Pigmenti - 30830										
492	4	A	78	24	F		y-			
1236	9	B	85	3	F	W	y-			

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY					
Leigh Encephalomyelopathy - 25600									
1503	2	A	45	3 1/2	F	W	--		Infantile subacute
Lowe Oculocerebrorenal Syndrome - 30900									
1676	2	B	107	10	M	W	y-		See Aging Repository
Marfan Syndrome - 15470									
35	4	A	26	12	M	W	+-(0)		Sib
36	4	A	26	10	M	W	+-(0)		Sib
Mental Retardation, X-Linked - 30950									
1228	3	C	66	2	M	W	y-		
Muscular Dystrophy									
Becker Type (Progressive Tardive) - 31010									
2298	8	K	202	18	M		y-		
Duchenne Type (Pseudohypertrophic Progressive) - 31020									
2339	8	A	88	20 wk.F	F	M	y-		
Myositis Ossificans Progressiva - 13510									
513	3	A	8	16	M	W	+-(0)		
783	3	A	107		M	B	+-(0)		
Neurofibromatosis (Von Recklinghausen Disease) - 16220									
622	3	A	95	8	M		+-(0)		Atypical; See GM-1634 Lymphoid
1633	3	B	104	61	M	W	+-(0)		Normal skin; other fibroblasts also available; see also GM-1641 Lymphoid
1639	3	B	104	19	F	B	+-(0)		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY					
Neurofibromatosis (Von Recklinghausen Disease), continued									
1860	6	B	104	41	M	W	+-(0)		Normal skin; other fibroblasts also available; see also GM-1861 Lymphoid
Osteogenesis Imperfecta - 16620									
744	8	C	57	1 da.	F	W	+-(0)		
1093	7	A	95	8	M	W	+-(0)		
1436	3	A	147	24	M	W	+-(0)		
Papular Mucinosis									Genetic or acquired
948	3	A	124	60	M	W			Gingiva
950	2	C	124	60	M	W			From skin of non-lesioned leg — Same patient
951	3	C	124	60	M	W			From skin of lesioned leg — Same patient
Potter Syndrome (Renal Agenesis) - 26670									
630	3	A	95	1 da.	M		--		
869	3	C	95	1 da.	F	W	--		
Progeria (See Aging Repository) - 26140									Multiplication rate slow
917	14	C	49	17	F		--		From Canadian Repository
989	11	A	55	20	M		--		Atypical
990	9	C	55		M		--		Atypical
991	8	A	55	4	M		--		
1177	8	C	49	9	M		--		From Canadian Repository
1178	18	C	49	34	M		--		From Canadian Repository
1972	8	B	91	14	F		--		

DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
Retinoblastoma (See Aging Repository) - 18020									
913	3	C	65	2	M	W	+?		Skin] Same
914	3	C	65	2	M	W	+?		Conjunctiva] patient
1879	2	B	155	11	F	W	+-(0)		Proband; patient also has osteosarcoma
1880	3	B	155	37	F	W	+-(0)		Mother
Schizophrenia and Psychiatric Disorders - 18150									
1792	3	B	137	26	M	W			Affected son; see GM-1793 Lymphoid
1833	4	B	137	56	M	W			Proband; atypical psychosis; see GM-1834 Lymphoid
1835	3	B	137	27	F	W			Affected daughter; see GM-1836 Lymphoid
1882	3	B	137	25	F	W			Normal daughter; see GM-1883 Lymphoid
1824	3	B	137	56	M	W			"Carrier" for schizo.; father of GM-1827 Lymphoid: see GM-1825 Lymphoid
1846	5	B	137	20	M	W			Normal first cousin of GM-1827 Lymphoid; see GM-1847 Lymphoid
1844	3	B	137	55	M	W			Affected father of GM-1846; see GM-1845 Lymphoid
Sea-Blue Histiocyte Disease - 26960									
843	3	C	66	10	F	W	--		Sib
844	3	C	66		M	W	--		Sib
1912	3	B	163	24	F	W	--		See GM-1913 Lymphoid
Spinocerebellar Ataxia									
1960	5	B	105	15	F	W			Culture grows slowly

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY					
Testicular Feminization Syndrome - 31370									X-Linked
1404	4	C	109	20	F	B	y-		46XY
1628	2	A	125	1	F	W	y-		46XY; proband
1948	3	B	125	27	F	W	+-(0)		Mother
1721	2	A	107	15	F	W	y-		46XY
2300	2	B	107	8 mo.	F	B	y-		46XY; skin — Same
2301	4	B	107	8 mo.	F	B	y-		46XY; gonad — patient
Thanatophoric Dwarfism - 27365									
1422	2	A	107	2 wk.	F	B	--		
Tuberous Sclerosis - 19110									
1643	3	B	104	17	M	B	+-(0)		Normal skin; other fibroblasts also available; see also GM-1636 Lymphoid
1644	2	A	104	20	F	B	+-(0)		Normal skin; other fibroblasts also available; see also GM-1638 Lymphoid
Werdnig-Hoffmann Disease									
232	2	A	78	7 mo.	M	W	--		
Wilson Disease (Hepatolenticular Degeneration) - 27790									
32	4	A	95	9	F	W	--		
33	4	A	95	16	M	W	--		
Wiskott-Aldrich Syndrome - 30100									
1598	2	A	107	4	M	W	y-		
Zellweger Syndrome - 21410									
228	3	A	107	2 mo.	M	W	--		

HUMAN FIBROBLAST CULTURES WITH CHROMOSOMAL ABERRATIONS

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE									
Ataxia-Telangiectasia - 20890									
367	3	A	107	17	M	W	--		Lung cells
647	7	C	133	17	M	W	--	A	
648	9	C	133	15	M	W	--	A	
1588	13	B	77		M	W	--	A	
1740	18	B	77		M	W	--		
1829	10	C	133	7	M	W	--		
1841	11	C	133	13	M		--		
1936	6	G	77	16	F	W	--		Sib
1937	9	G	77	24	F	W	--		Sib
1970	2	B	155	3	M	W	--		Proband
1977	3	B	155	30	M	W	+-(O)		Father
1986	2	B	155	28	F	W	+-(O)		Mother
2052	3	B	77	15	F	W	--		
Bloom Syndrome - 21090									
811	17	N	44	4	M	W	--	A	
1492	11	B	77		M	W	--		
1493	10	B	77	3	M	W	--		
1620	22	C	44	8 mo.	F	W	--		See Aging Repository
Fanconi Anemia (Pancytopenia) - 22765									
368	6	J	133	8	M	B	--	A	
369	11	A	133	6	M	W	--		
391	6	J	133	6	F	W	--		
646	7	C	133	16	F	W	--		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
			SYNDROMES WITH INCREASED CHROMOSOME BREAKAGE						
Fanconi Anemia (Pancytopenia), continued									
1309	3	C	133	12	M	B	--		
449	10	J	44	6	F	W	--	A	
1746	5	A	77	15	M	W	--		
2053	4	B	77	12	F	W	--		
2061	8	A	133	20	F	W	--		
2361	8	K	188	14	M		--		
2362	11	K	188	14	M		--		
2363	10	K	188	15	M		--		
2364	12	K	188	24	M		--		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-	Remarks
							TRANSLOCATIONS			
Chromosome 1										
97*	3	A	107	1	F	W	46,X,t(X;1)(Xpter ➤Xq26 ::1q12 ➤1qter;1pter ➤1q12::Xq26 ➤Xqter)	B	A	De novo translocation, Beckwith-Wiedemann Syndrome
860	11	B	36	13	F	W	46,XX,der(5),ins(5;1)(q13;q25q32)pat	U	A	Proband; grows slowly
861	7	B	36	40	M	W	46,XY,ins(5;1)(5pter ➤5q13::1q25 ➤1q32::5q13 ➤5qter;1pter ➤1q25::1q32 ➤1qter)	B	A	Father
1421	12	A	109	53	M	B	46,XY,t(1;6)(q42;q21)	B	A	
126*	4	A	56	33	M	W	46,XY,t(1;15)(1qter ➤1p36::15q1 ➤15qter; 15pter ➤15q1::1p36 ➤1pter)	B	A	
201*	10	A	30	13	F	W	47,XX,+21,t(1;17)(1pter ➤1p32::17p13 ➤17qter; 17pter ➤17p13::1p32 ➤1qter)	B	A	
1356	2	A	66	26	F	W	46,XX,t(1;7)(1pter ➤1p34::7p13 ➤7qter;7pter ➤7p13::1p34 ➤1qter)	B	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 7; 11; 10

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 1, continued										
1229	3	B	7	3	F	B	46,XX,-1,+der(1), t(1;2)(p36;q31)pat	U	A	Proband
1230	2	B	7	44	M	B	46,XY,t(1;2)(1qter➤1p36::2q31➤2qter; 2pter➤2q31::1p36➤1pter)	B	A	Father
1550	6	A	140	31	M	W	46,XY,t(1;3;4)(1pter➤1q32::3p21➤3pter; 1qter➤1q32::3p21➤3q29::4p14➤4pter; 4qter➤4p14::3q29➤3qter)	B	A	
257*	3	A	2	26	F		46,XX,t(1;2)(1pter➤1q32::2p23➤2pter; 1qter➤1q32::2p23➤2qter)	B	A	
1564	9	C	151	28	F	W	45,XX,t(1;21)(q42or43;q11)			
1813	3	B	66	19	F	W	46,XX,t(1;21)(q12;q22)pat/46,XX,t(1;21)(q12;q22)pat,t(X;15)(p11;q13)	U	A	Menstrual dysfunction; Adj-2 segregation

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #18

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 1, continued										
1881	7	A	140	3	M	W	46,XY,t(1;21)(1qter➤1p32::21q22➤21qter; 21pter➤21q22::1p32➤1pter)	U	A	Suspected Sanfilippo
Chromosome 2										
257*	3	A	2	26	F		46,XX,t(1;2)(q32;p23)		A	t(1;2); see Chromosome 1
1229	3	B	7	3	F	B	46,XX,-1,+der(1), t(1;2)(p36;q31)pat	U	A	Proband; t(1;2); see Chromosome 1
1230	2	B	7	44	M	B	46,XY,t(1;2)(p36;q31)	B	A	Father; t(1;2); see Chromosome 1
501*	3	A	14	4 1/2	M	W	46,XY,der(2),t(2;4)(2qter➤2p25::4q21➤4qter)pat	U	A	Proband
1064*	7	A	14	39	M	W	46,XY,t(2;4)(2qter➤2p25::4q21➤4qter; 4pter➤4q21::2p25➤2pter)	B	A	Father
327*	4	A	141	24	M	W	46,XY,t(2;8)(2pter➤2q13::8q24➤8qter; 8pter➤8q24::2q13➤2qter)	B	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 18; 38; 39; 44

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 2, continued										
845	7	C	18	18	F	W	46,XX,t(2;8)(2pter➤2q37::8q13➤8qter; 8pter➤8q13::2q37➤2qter)mat	B	A	Proband; primary Amenorrhea
846	8	C	18	54	F	W	46,XX,t(2;8)(2pter➤2q37::8q13➤8qter; 8pter➤8q13::2q37➤2qter)	B	A	Mother; multiple spontaneous abortions
1225	10	B	77	3 mo.	F	I	46,XX,t(2;20)(2qter➤2p21::20p13➤20pter; 20qter➤20p13::2p21➤2pter)		A	Indian (India); de novo; has multiple congenital anomalies
692	5	A	1	8	M	W	45,XY,-21,t(2;21)(q3;q22)mat	U	B	Proband; mentally retarded
693	5	A	1	27	F	W	46,XX,t(2;21)(q3;q22)	B	B	Mother
1579	8	A	30	35	F	W	46,XX,t(2;13)(p1;q34)		B	
1848	3	B	179	35	M	W	46,XY,der(10),t(2;10)(p24;q26)mat	U	B	Cousin
1849	4	B	179	26	M	W	46,XY,der(10),t(2;10)(2pter➤2p24::10q26➤10pter)mat	U	A	Cousin
1683	3	B	179	28	F	W	46,XX,t(2;10)(p24;q26)	B	A	Proband; cousin

Possible red cell acid phosphatase variant

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
							TRANSLOCATIONS			
Chromosome 3										
1550	6	A	140	31	M	W	46,XY,t(1;3;4)(q32; p21q29;p14)	B	A	t(1;3;4); see Chromosome 1
194*	15	A	30	19	F	W	46,XX,t(X;3)(Xpter➤Xq2 ::3q1➤3qter;3pter➤3q1 ::Xq2➤Xqter)	B	A	Possibly reciprocal; 3q12 or 3q13 break; break pt. on X distal to Xq25
Chromosome 4										
1550	6	A	140	31	M	W	46,XY,t(1;3;4)(q32; p21q29;p14)	B	A	t(1;3;4); see Chromosome 1
501*	3	A	14	4 1/2	M	W	46,XY,der(2),t(2;4) (p25;q21)pat	U	A	t(2;4); see Chromosome 2; Proband
1064*	7	A	14	39	M	W	46,XY,t(2;4)(p25;q21)	B	A	t(2;4); see Chromosome 2; Father
1087	3	A	66	2 mo.	F		46,XX,t(4;5)(p;q)			
773*	5	C	66	26	F		46,XX,t(4;7)(4qter➤ 4p16::7q34➤7qter; 7pter➤7q34::4p16➤ 4pter)	B	A	Mother
1220*	3	A	66	2	F		46,XX,der(4),t(4;7) (4qter➤4p16::7q34➤ 7qter)mat	U	A	Proband

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 29; 38; 39; 47; 49

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 4, continued										
157*	11	A	30	28	M	W	49,XXXXY,t(4;11)(4pter➤4q35::11q23➤11qter;11pter➤11q23::4q35➤4qter)pat	B	A	Severe mental retardation, sexual infantilism
380*	2	A	78	35	F		46,XX,rcp(4;11)(4pter➤4q25::11q13➤11qter;11pter➤11q13::4q25➤4qter)	B	A	
1101	7	C	7	32	M	W	46,XY,t(4;12)(p1;p11)	B		
972	3	C	67	12 1/2	F	B	46,XX,t(4;13)(4pter➤4q31::13q14➤13qter;13pter➤13q14::4q31➤4qter)	B	A	
624*	2	A	77	38	F	W	46,XX,t(4;15)(4pter➤4q11::15p11➤15pter;15qter➤15p11::4q11➤4qter)	B	A	
98*	5	A	119	10 mo.	M		46,XY,der(21),t(4;21)(p11;p12)mat	U	A	
1091	3	A	58	27	F	W	46,XX,t(4;10)(q32;q22)	B	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 23; 16; 22; 17

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 5										
860	11	B	36	13	F	W	46,XX,der(5),ins(5;1)(q15;q32q25)pat	U	A	t(1;5); grows slowly; see Chromosome 1; Proband
861	7	B	36	40	M	W	46,XY,ins(5;1)(q15;q32q25)	B	A	t(1;5); see Chromosome 1 Father
1087	3	A	66	2 mo.	F		46,XX,t(4;5)(p;q)			t(4;5); see Chromosome 4
1221	6	C	21	1 1/2	M	B	46,XY,-5,+ins(5;6)(q33;q15q27)mat	U	B	Partial trisomy of 6q15q27; Proband
1222	5	C	21	23	F	B	46,XX,ins(5;6)(q33;q15q27)mat	B	B	Mother
1524	3	C	21	19	M	B	46,XY,-5,+ins(5;6)(q33;q15q27)		B	Cousin
71*	15	A	15	19	F	W	46,XX,-5,der(5),t(5;?)(p1;?)mat		A	See Deletions Cri du Chat
589*	2	A	12	17 1/2	M	W	46,XY,t(5;14)(5qter➤5p14::14q21➤14qter;14pter➤14q21::5p14➤5pter)		A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 36; 41

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 5, continued										
344*	4	A	66	6 1/2	M	W	45,XY,del(5)(pter ➤p14:),t(5;15)(5qter ➤5p14::15q1➤15qter), t(9:11)(9qter➤9p24:: 11q23➤11qter;11pter➤ 11q23::9p24➤9pter)	U	A	Cri du Chat
1535	6	A	2	16	F	B	46,XX,der(12),t(5;12) (q31:q24)mat,9qh+,21s+	U		Proband
1536	6	C	2	38	F	B	46,XX,t(5;12)(q31;q24), 9qh+,21s+	B		Mother
1678	3	B	140	33	F		46,XX,t(5;10)(p15;p13)	B	B	
Chromosome 6										
1421	12	A	109	53	M	B	46,XY,t(1;6)(q43;q21)	B	A	t(1;6); see Chromosome 1
1137	6	D	17	1 mo.	M	B	48,XXY,+21,rcp(6;10) (6qter➤6p22 or 24:: 10p12➤10pter;10qter➤ 10p12::6p22 or 24➤6pter)		B	
1524	3	C	21	19	M	B	46,XY,-5,+ins(5;6) (q33;q15q27)mat	U	B	t(5;6); Cousin; see Chromosome 5
1221	6	C	21	1 1/2	M	B	46,XY,-5,+ins(5;6) (q33;q15q27)mat	U	B	t(5;6); Proband; see Chromosome 5
1222	5	C	21	23	F	B	46,XX,ins(5;6)(q33; q15q27)mat	B	B	t(5;6); Mother see Chromosome 5

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #27

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 6, continued										
144*	5	A	12	34	F	W	46,XX,der(6),t(6;21)(21qter ► 21q11::6p25 ► 6qter)mat	U	A	13% of cells found to be balanced in passage 1 after recovery; these tend to increase with passage in culture
610*	7	A	75	3 mo.	F	W	46,XX,t(6;18)(6pter ► 6q21::18p11 ► 18pter; 6qter ► 6q21::18p11 ► 18qter)	B	A	
1605	3	A	166	34	M	W	46,XY,t(6;11)(6qter ► 6p2::11q23 ► 11qter; 11pter ► 11q23::6p2 ► 6pter)	B	B	
2068	5	J	88	4	F	W	46,XX,t(6;7)(6pter ► 6q27::7q22 ► 7qter; 7pter ► 7q22::6q27 ► 6qter)	B	A	
1139	4	A	95	21	F	W	46,XX,t(15;17)(q15;p13),t(6;13)(p21;q34)		A	Culture is mosaic 64% carry only the t(15;17) 36% carry both
Chromosome 7										
1356	2	A	66	26	F	W	46,XX,t(1;7)(p34;p13)	B	A	t(1;7); see Chromosome 1

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 6; 28

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 7, continued										
773	5	C	66	26	F		46,XX,t(4;7)(p16;q34)	B	A	t(4;7); see Chromosome 4; Mother
1220	3	A	66	2	F		46,XX,der(4),t(4;7)(p16;q34)mat	U	A	t(4;7); see Chromosome 4; Proband
44*	5	A	97	6	M	W	46,XY,t(7;10)(7qter►7p2::10q11►10qter;10pter►10q11::7p2►7pter)	B	A	Congenital malformations, severe retardation although apparently balanced
1696	9	C	176	30	F	W	46,X,t(X;7)(Xqter►Xq21::7p22►7qter;Xpter►Xq21::7p22►7pter)	B	B	
2068	5	J	88	4	F	W	46,XX,t(6;7)(q27;q22)	B	A	t(6;7); see Chromosome 6
657	3	A	107	4 wk.	M	W	46,XY,t(7;18)(7pter►7q36::18q21►18qter;18pter►18q21::7q36►7qter)	U	A	
Chromosome 8										
845	7	C	18	18	F	W	46,XX,t(2;8)(q37;q13)mat	B	A	t(2;8); Proband; see Chromosome 2
846	8	C	18	54	F	W	46,XX,t(2;8)(q37;q13)	B	A	t(2;8); Mother; see Chromosome 2
327	4	A	141	24	M	W	46,XY,t(2;8)(q13;q24)	B	A	t(2;8); see Chromosome 2

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #12

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 8, continued										
1512	6	A	30	26	M	W	46,XY,t(8;10)(p21;p15)	B	B	
213*	5	A	82	35	M	W	46,XY,t(8;12)(8pter►8p23::12p11►12qter; 12pter►12p11::8p23►8qter)	B	A	
Chromosome 9										
1387	4	A	61	14	F	W	46,XX,-13,+der(9), t(9;13)(q22;q12)mat	U	B	Result of adj-2 seg.
988*	10	G	82	3 wk.	M	W	46,XY,t(9;17)(9qter►9p13::17q25►17qter; 17pter►17q25::9p13►9pter)	B	A	Multiple congenital anomalies; 13% of cells are polyploid
705	13	B	16	10 mo.	F	W	46,X,t(X;9)(q12;p24)	B	B	Grows slowly
1414	5	C	68	28	F	B	46,X,-X,+der(9),rcp (X;9)(q11;q32)mat	U	B	Proband; Turner's Syndrome
1429	8	A	68	55	F	B	47,X,+10,rcp(X;9) (q13;q34)	B	A	Mother; mosaic, 14% do not show +10
1664	6	C	113	40	M	W	46,XY,t(9;13)(q13;q12)	B	B	See Chromosome 13
1734	3	B	140	40	M		46,XY,der(15),t(9;15) (p11;q11)mat	U	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 8; 42

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 9, continued										
1750	2	B	70	2 1/2	M	W	47,XY,+der(14)t(9;14)(p24;q22)mat	U	B	Proband
1751	2	B	70	26	F	W	46,XX,t(9;14)(p24;q22)	B	B	Mother see Chromosome 14
1892	5	B	132	37	F	W	46,XX,rcp(9;18)(p24;q12)	B	B	
Chromosome 10										
1137	6	D	17	1 mo.	M	B	48,XXY,+21,rcp(6;10)(p22 or 24;p12)		B	t(6;10); see Chromosome 6
44*	5	A	97	6	M	W	46,XY,t(7;10)(p2;q11)	B	A	t(7;10); see Chromosome 7
1512	6	A	30	26	M	W	46,XY,t(8;10)(p21;p15)	B	B	t(8;10); see Chromosome 8
1396	3	A	95	3	M	W	46,XY,der(10),t(10;16)(q26;q22)pat		A	
216*	4	A	143	29	M	W	46,XY,t(10;17)(10pter→10q24::17p13→17pter;17qter→17p13::10q24→10qter)	B	A	Father
217*	6	A	143	9 da.	M	W	46,XY,-17,+der(17),t(10;17)(17qter→17p13::10q24→10qter)pat	U	A	Proband, deceased; culture grows slowly

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 12; 13; 14

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 10, continued										
959*	6	C	143	11	M	W	same karyotype as GM-217, pat	U	A	Paternal fourth cousin of GM-217
1413	8	B	132	1 1/2	F	B	46,XX,der(10),rcp(10;21)(q26;q21)mat	U	B	Proband
1580	6	B	132	48	F	B	46,XX,rcp(10;21)(q26;q21)	B	B	Grandmother of proband; aunt of GM-1399
1399	5	B	132	20	M	B	47,XY,+der(21),rcp(10;21)(q26;q21)mat	U	B	Nephew of GM-1580; Cousin of GM-1413
984	4	C	95	34	F	W	46,XX,t(10;21)(q24;q22)	B		Mother
983	3	C	95	14 mo.	M	W	46,XY,der(21),t(10;21)(q24;q22)mat	U		Proband
1678	3	B	140	33	F		46,XX,t(5;10)(p15;p13)			See Chromosome 5
1848	3	B	179	35	M	W	46,XY,der(10),t(2;10)(p24;q26)mat	U	B	See Chromosome 2 Cousin
1849	4	B	179	26	M	W	46,XY,der(10),t(2;10)(p24;q26)mat	U	A	See Chromosome 2 Cousin
1683	3	B	179	28	F	W	46,XX,t(2;10)(p24;q26)	B	A	See Chromosome 2 proband

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #37

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 10, continued										
1091	3	A	58	27	F	W	46,XX,t(4;10)(q32;q22)	B	B	
Chromosome 11										
157*	11	A	30	28	M	W	49,XXXXY,t(4;11)(q35;q23)pat	B	A	t(4;11); see Chromosome 4
380*	2	A	78	35	F		46,XX,rcp(4;11)(q25;q13)	B	A	t(4;11); see Chromosome 4
980	2	A	12	33	F	W	45,XX,-22,t(11;22)(11pter➤11q25::22q11➤22qter)	U	B	
1605	3	A	166	34	M	W	46,XY,t(6;11)(6qter➤6p2::11q23➤11qter;11pter➤11q23::6p2➤6pter)	B	B	t(6;11); see Chromosome 6
Chromosome 12										
1101	7	C	7	32	M	W	46,XY,t(4;12)(p1;p11)	B		t(4;12); see Chromosome 4
1535	6	A	2	16	F	B	46,XX,der(12),t(5;12)mat, 9qh+,21s+	U	B	Proband; t(5;12); see Chromosome 5
1536	6	C	2	38	F	B	46,XX,t(5;12), 9qh+,21s+	B	B	Mother; t(5;12); see Chromosome 5
213*	5	A	82	35	M	W	46,XY,t(8;12)(p23;p11)	B	A	t(8;12); see Chromosome 8
1665	3	B	140	28	F	W	46,XX,t(12;21)(12p21q)	B	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 23; 16; 8

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 13										
1579	8	A	30	35	F	W	46,XX,t(2;13)(p1;q34)		B	t(2;13); see Chromosome 2
972	3	C	67	12 1/2	F	B	46,XX,t(4;13)(q31;q14)	B	B	t(4;13); see Chromosome 4
1387	4	A	61	14	F	W	46,XX,-13,+der(9), t(9;13)(q22;q12)mat	U	B	t(9;13); see Chromosome 9
1555	5	C	113	13	M	W	47,XY,+der(13),t(13;17) (q14;p13)mat	U	B	Sib
1663	13	A	113	14	M	W	46,XY,+der(17),t(13;17) (q14;p13)mat	U	B	Proband; partial trisomy 13
85	5	A	143	2 wk.	M	B	46,XY,-13,+t(13q13q)	U	A	Clinical Trisomy 13
1296	4	C	12	47	F	W	45,XX,t(13q15q)	B		
1224	7	B	77	3 mo.	F	W	46,XX,der(13),t(13;18) (q32;q11)pat	U	A	
392*	9	A	2	1	F	W	45,XX,t(13;22)(13qter►cen►22qter)	B	A	
627*	3	A	58	30	F	B	46,XX,t(13;22)(13pter►13q22::22q13►22qter; 22pter►22q13::13q22►13qter)	B	A	
1664	6	C	113	40	M	W	46,XY,t(9;13)(q13;q12)	B	B	See Chromosome 9

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 19; 30

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 13, continued										
2018	12	B	77	2	F	W	45,XX,tan(13;13)(pter ►q34::q12►qter)	U	A	
1139	4	A	95	21	F	W	46,XX,t(15;17)/46,XX, t(15;17),t(6;13)		A	t(15;17);t(6;13) see Chromosome 6
Chromosome 14										
589*	2	A	12	17 1/2	M	W	46,XY,t(5;14)(p14;q21)		A	t(5;14); see Chromosome 5
479*	12	C	48	35	F	W	45,XX,t(14;15)(14qter► cen►15qter)		A	
981	3	C	148	4	F	B	46,XX,der(14),t(14; 20)(p11;p11),inv(9) (pter►p13::q13►p13:: q13►qter)	U	A	Proband
982	3	C	148	Adult	F	B	46,XX,t(14;20)(p11;p11)	B	A	Mother
5*	5	A	111	30	F	W	45,XX,t(14;22)(14qter► cen►22qter)		A	
73*	5	C	111	Adult	F	W	46,X,t(X;14)(Xpter► Xq13::14qter;14pter► 14q32::Xq13►Xqter)	B	A	Mother
74*	10	S	111	23	M	W	47,Y,+der(X),+der(14), +der(14),t(X;14)(q13; q32)mat	U	A	Proband; Klinefelter Syndrome

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 41; 33; 3; 1; 2

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 14, continued										
1750	2	B	70	2 1/2	M	W	47,XY,+der(14),t(9;14)(p24;q22)mat	U	B	Proband
1751	2	B	174	26	F	W	46,XX,t(9;14)(p24;q22)	B	B	Mother; t(9;14); see Chromosome 9
2044	2	B	164	6 1/2	M		45,XY,-16,t(14;16)(16;18)(14qter ➤ 14p12::16p11 ➤ 16pter;18qter ➤ 18p1::16q12 ➤ 16qter)	U	A	
Chromosome 15										
126*	4	A	56	33	M	W	46,XY,t(1;15)(p36;q1)	B	A	t(1;15); see Chromosome 1
624*	2	A	77	38	F	W	46,XX,t(4;15)(q11;p11)	B	A	t(4;15); see Chromosome 4
344*	4	A	66	6 1/2	M	W	45,XY,del(5),t(5;15)(p14;q1),t(9;11)(p24;q23)	U	A	t(5;15)+t(9;11); see Chromosome 5
1296	4	C	12	47	F	W	45,XX,t(13q15q)	B		t(13;15); see Chromosome 13
479	12	C	48	35	F	W	45,XX,t(14q15q)		A	t(14;15); see Chromosome 14
1139	4	A	95	21	F	W	46,XX,t(15;17)(q22;p13)/46,XX,t(15;17),t(6;13)	B	A	t(15;17)t(6;13); see Chromosome 6
17*	4	A	12	2 1/2	M	W	45,XY,t(15;18)(15qter ➤ 15q1::18q23 ➤ 18pter)	U	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 11; 22; 27; 4

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 15, continued										
118*	5	A	52	29	F	W	46,XX,-15,+der(Y), t(Y;15)(Yqter ► Yq11:: 15p1 ► 15qter)pat	U	A	Uterine tissue
1734	3	B	140		M		46,XY,der(15),t(9;15) (p11;q11)mat	U	B	t(9;15); see Chromosome 9
1813	3	B	66	19	F	W	46,XX,t(1;21)(q12;q22) pat/46,XX,t(1;21)(q12; q22)pat,t(X;15)(p11;q13)	U	A	See Chromosome 1
Chromosome 16										
1396	3	A	95	3	M	W	46,XX,der(10),t(10;16) (q26;q22)pat		A	t(10;16); see Chromosome 10
2044	2	B	164	6 1/2	M		45,XY,-16,t(14;16) (16;18)(p12;p11q12;p1)	U	A	t(14;16)t(16;18); see Chromosome 14
Chromosome 17										
201*	10	A	30	13	F	W	47,XX,+21,t(1;17) (p32;p13)	B	A	t(1;17); see Chromosome 1

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 31; 10

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 17, continued										
216*	4	A	143	29	M	W	46,XY,t(10;17)(q24;p13)	B	A	t(10;17); Father see Chromosome 10
217*	6	A	143	9 da.	M	W	46,XY,-17,+der(17), t(10;17)(q24;p13)pat	U	A	t(10;17); Proband see Chromosome 10
959	6	C	143	11	M	W	46,XY,-17,+der(17), t(10;17)(q24;p13)pat	U	A	t(10;17); pat 4th cousin; see Chromosome 10
1555	5	C	113	13	M	W	47,XY,+der(13),t(13;17)(q14;p13)mat	U	B	t(13;17); Sib; see Chromosome 13
1663	13	A	113	14	M	W	46,XY,-17,+der(17), t(13;17)(q14;p13)mat	U	B	t(13;17); Proband; see Chromosome 13
1139	4	A	95	21	F	W	46,XX,t(15;17)(q22;p13)/46,XX,t(15;17),t(6;13)	B	A	t(15;17); t(6;13) see Chromosome 6
271*	3	A	58	28	F	W	46,XX,t(17;19)(17pter→17q23::19p13→19pter; 19qter→19p13::17q23→17qter)	B	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 13; 14; 37; 15

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 17, continued										
119*	4	A	87	31	F	W	46,XX,t(17;22)(17qter➤17p13::22q11➤22qter; 22pter➤22q11::17p13➤17pter)	B	A	
988*	10	G	82	3 wk.	M	W	46,XY,t(9;17)(p13;q25)	B	A	t(9;17); see Chromosome 9
Chromosome 18										
610*	7	A	75	3 mo.	F	W	46,XX,t(6;18)(q21;p11)	B	A	t(6;18); see Chromosome 6
1224	7	B	77	3 mo.	F	W	46,XX,der(13),t(13;18)(q32;q11)pat	U	A	t(13;18); see Chromosome 13
17*	4	A	12	2 1/2	M	W	45,XY,t(15;18)(q1;q23)	U	A	t(15;18); see Chromosome 15
657	3	A	107	4 wk.	M	W	46,XY,t(7;18)(q36;q21)	U	A	t(7;18); see Chromosome 7
1892	5	B	132	37	F	W	46,XX,rcp(9;18)(p24;q12)	B	B	t(9;18) see Chromosome 9
2044	2	B	164	6 1/2	M		45,XY,-16,t(14;16)(16;18)(p12;p11q12;p1)	U	A	t(14;16)t(16;18); see Chromosome 14
Chromosome 19										
271*	3	A	58	28	F	W	46,XX,t(17;19)(q23;p13)	B	A	t(17;19); see Chromosome 17

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 9; 42; 28; 4; 15

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified		Remarks
							TRANSLOCATIONS			
Chromosome 19, continued										
89	5	A	43	Adult	F	W	46,X,t(X;19)(Xpter➤Xq2::19q13or19p13➤19qter or 19pter?; 19pter or 19qter➤19p13 or19q13::Xq2➤Xqter)	B	A	
Chromosome 20										
1225	10	B	77	3 mo.	F	I	46,XX,t(2;20)(p21;p13)		A	t(2;20); see Chromosome 2
981	3	C	148	4	F	B	46,XX,der(14),t(14;20)(p11;p11)mat,inv(9)(p13q13)	U	A	Proband; t(14;20); see Chromosome 14
982	3	C	148	Adult	F	B	46,XX,t(14;20)(p11;p11)	B	A	Mother; t(14;20); see Chromosome 14
Chromosome 21										
1564	9	C	151	28	F		45,XX,t(1;21)(q42or43;q11)			t(1;21); see Chromosome 1
692	5	A	1	8	M	W	45,XY,-21,t(2;21)(q3;q22)mat	U	B	t(2;21); Proband; see Chromosome 2
693	5	A	1	27	F	W	46,XX,t(2;21)(q3;q22)	B	B	t(2;21); Mother; see Chromosome 2
98*	5	A	119	10 mo.	M		46,XY,der(21),t(4;21)(p11;p12)mat	U	A	t(4;21); see Chromosome 4
144*	5	A	12	34	F	W	46,XX,der(6),t(6;21)(p25;q11)mat	U	A	t(6;21); see Chromosome 6

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 17; 6

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
							TRANSLOCATIONS			
Chromosome 21, continued										
1413	8	B	132	1 1/2	F	B	46,XX,der(10),rcp(10;21)(q26;q21)mat	U	B	t(10;21); Proband; see Chromosome 10
1580	6	B	132	38	F	B	46,XX,rcp(10;21)(q26;q21)	B	B	t(10;21); Grandmother; see Chromosome 10
1399	5	B	132	20	M	B	47,XY,+der(21),rcp(10;21)(q26;q21)mat	U	B	t(10;21); Cousin; see Chromosome 10
1581	7	B	132	25	F	B	46,X,t(X;21)(q11;p11?)		B	Mother
1730	11	C	132	5	F	B	46,XX,der(21),t(X;21)(q11;p11?)mat		B	Proband
1665	3	B	140	28	F	W	46,XX,t(12;21)(12p21q)	B	B	t(12;21); see Chromosome 12
1700	3	B	95	6	M		46,XY,t(21;22)(p12;q11)	B	B	
1881	7	A	140	3	M		46,XY,t(1;21)(p32;q22)		A	t(1;21); suspected Sanfilippo; see Chromosome 1
2058	5	B	165	11 da.	F	W	46,XX,/46,XX,t(21;21)	U	B	87%/13% at passage 8; see Trisomy 21, Down's syndrome
1813	3	B	66	19	F	W	46,XX,t(1;21)(q12;q22)pat/46,XX,t(1;21)(q12;q22)pat,t(X;15)(p11;q13)	U	A	See Chromosome 1

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome 22										
980	2	A	12	33	F	W	45,XX,-22,t(11;22)(q25;q11)	U	B	t(11;22); see Chromosome 11
392*	9	A	2	1	F	W	45,XX,t(13;22)(13q22q)	B	A	t(13;22); see Chromosome 13
627*	3	A	58	30	F	B	46,XX,t(13;22)(q22;q13)	B	A	t(13;22); see Chromosome 13
5*	5	A	111	30	F	W	45,XX,t(14;22)(14q22q)		A	t(14;22); see Chromosome 14
119*	4	A	87	31	F	W	46,XX,t(17;22)(p13;q11)	B	A	t(17;22); see Chromosome 17
1700	3	B	95	6	M		46,XY,t(21;22)(p12;q11)	B	B	t(21;22); see Chromosome 21
Chromosome X										
97*	3	A	107	1	F	W	46,X,t(X;1)(q26;q12)	B	A	t(X;1); see Chromosome 1
194*	15	A	30	19	F	W	46,XX,t(X;3)(q2;q1)	B	A	t(X;3); see Chromosome 3
705	13	B	16	10 mo.	F	W	46,X,t(X;9)(q12;p24)	B	B	t(X;9); see Chromosome 9
1414	5	C	68	28	F	B	46,X,-X,+der(9),rcp(X;9)(q11;q32)mat	U	B	t(X;9); Proband; see Chromosome 9
1429	8	A	68	55	F	B	47,X,+10,rcp(X;9)(q13;q34)	B	A	t(X;9); Mosaic Mother; see Chromosome 9

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 19; 30; 3; 9; 7; 29

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Verified	Remarks
							TRANSLOCATIONS			
Chromosome X, continued										
73*	5	C	111	Adult	F	W	46,X,t(X;14)(q13;q32)	B	A	t(X;14); Mother; see Chromosome 14
74*	10	S	111	23	M	W	47,Y,+der(14),t(X;14)(q13;q32)mat	U	A	t(X;14); Proband; see Chromosome 14
89	5	A	43	Adult	F	W	46,X,t(X;19)(q2;q13or p13)	B	A	t(X;19); see Chromosome 19
1581	4 IMR	B	132	25	F	B	46,X,t(X;21)(q11;p11?)		B	t(X;21); Mother; see Chromosome 21
1730	11	C	132	5	F	B	46,XX,der(21),t(X;21)(q11;p11?)mat		B	t(X;21); Proband; see Chromosome 21
1696	9	C	176	30	F	W	46,X,t(X;7)(q21;p22)	B	B	t(X;7); see Chromosome 7
2103	6	C	109	19	F	W	46,X,-Y,t(X;Y)(Yqter➤Yq11::Xp11➤Xq22)	U	B	2° Amenorrhea; see Chromosome Y
1813	3	B	66	19	F	W	46,XX,t(1;21)(q12;q22)pat/46,XX,t(1;21)(q12;q22)pat,t(X;15)(p11;q13)	U	A	See Chromosome 1
Chromosome Y										
118*	5	A	52	29	F	W	46,XX,-15,+der(Y),t(Y;15)(q11;p1)	U	A	t(Y;15); see Chromosome 15
2103	6	C	109	19	F	W	46,X,-Y,t(X;Y)(q11;p11)	U	B	t(X;Y); see Chromosome X

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 1; 2; 31

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
							INVERSIONS			
Chromosome 2										
735	17	A	77	17	F	W	46,Xdic,Xqi,inv(2)(p15q21)mat	B	B	Turner Syndrome
Chromosome 3										
1252	3	C	106	22	F	B	46,XX,inv(3)(p25q25),inv(9)(p11q13)	B	A	Mother; see Chromosome 9
1253	3	C	106	4 mo.	M	B	46,XY,rec(3),dup q,inv(3)(p25q25)mat	U	A	Proband; see Chromosome 9; Father is XYY (GM-1250)
Chromosome 4										
413	4	B	56	1	M		46,XY,inv(4)(pter ➤p14::q22 ➤p14::q22 ➤pter)mat	B	B	Proband; has minor malformations
414	4	B	56	23	F		46,XX,inv(4)(pter ➤p14::q22 ➤p14::q22 ➤pter)mat	B	B	Mother
Chromosome 9										
445	7	A	30	26	M	W	46,XY,inv(9)(p13q13)		B	Father; clinically normal
447	7	A	30	1	M	W	46,XY,inv(9)(p13q13)		B	Proband; congenital malformations
450	7	A	30	33	M	W	46,XY,inv(9)(p13q13)	B	B	Same patient; clinically normal
451	7	A	30	33	M	W	46,XY,inv(9)(p13q13)	B	B	Same patient; testicular tissue

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Bal. Unbal.	Veri-fied	Remarks
							INVERSIONS			
Chromosome 9, continued										
453	7	A	30	24	F	W	46,XX,inv(9)(p13q13)	B	B	Clinically normal
1251	2	C	106	4	M	B	46,XY,inv(9)(p11q13) mat	B	A	Half brother of proband
1252	3	C	106	22	F	B	46,XX,inv(3)(p25q25), inv(9)(p11q13)	B	A	Mother of proband & sib; see Inversion Chromosome 3
1253	3	C	106	4 mo.	M	B	46,XY,rec(3),dup q, inv(3)(p25q25)mat	U	A	Proband; father is XYY (GM-1250); see Inversion Chromosome 3
1918	6	B	61	28	M	B	47,XYqs,+21,inv(9) (p13q21)			See Trisomy, Chromosome 21
1920	2	B	61	23	M	B	47,XY,+21,inv(9)(p13q13) mat			See Trisomy, Chromosome 21
981	3	C	148	4	F	B	46,XX,der(14)t(14;20) (p11;p11)inv(9)(p13q13)	U	A	See t(14;20)
Chromosome 10										
446	7	A	30	26	M	W	46,XY,inv(10)(p11q11)	B	B	Clinically normal
448	7	A	30	1	M	W	46,XY,inv(10)(p11q21) pat	B	B	Proband; congenital malformations;
452	7	A	30	22	M	W	46,XY,inv(10)(p11q21)	B	B	Clinically normal; father
Chromosome 13										
437	3	A	66	32	F	W	46,XX,inv(13)(p13q21)	B	B	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
							INVERSIONS		
Chromosome 13, continued									
1570	2	A	119	5 mo.F	M		46,XY,rec(13)dup p,inv (13)(p12q32)		Paternal karyotype is 46,XY,inv(13) (p12q32)

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
							MONOSOMIES		
Chromosome 21									
137	4	A	75	5 1/2	M	W	45,XY,-21	A	
230	9	J	88	14 mo.	F	W	45,XX,-21	A	
Chromosome X									
775	4	C	107	13	F		45,X	A	
993	2	A	107	9	F	B	45,X	A	
1176	2	A	107	8	F	W	45,X	B	
857	#	A	95	1 da.	F	W	45,X	B	
563	4	A	66	14	F	B	45,X	B	Same patient; only XO cells found; tissue from right gonad
562	4	A	66	14	F	B	45,X/46,XX	B	Same patient; 4 cells, 46XX/46 cells, 45XO; tissue from left gonad
339	12	A	30	25	F	W	45,X/46,X,i(Xq)		See iso-X
314	16	A	30	60	F	W	45,X/46,X,dup(X)(qter ➤pter::pter ➤qter)	B	Ovarian dysgenesis without Turner stigmata
1441	2	A	74	3 1/2 mo.	M	W	45,X/46,X,iso(Y)		See iso-Y; tissue from right testis
978	7	C	143	15	F	W	46,XY/45,X		Karyotype based on leukocytes, tissue from right gonad; see Gonadal Dysgenesis
1723	2	B	203	23	F	W	45,X	A	

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
					TRISOMY/POLYSOMY				
Chromosome 8									
496	3	A	56	2	M		47,XY,+8/46,XY		2 cell lines in lymphocytes; 83% have +8; in skin fibroblasts 81% have +8
2030	7	A	30	23	M	W	46,XY/47,XY,+8	B	5%/95% in fibroblasts; mental retardation
Chromosome 9									
2329	6	J	39	18 da.	F	W	47,XX,+9/46,XX	B	10% of fibroblasts show the +9
Chromosome 10									
1429	8	A	68	55	F	B	47,X,+10,rcp(X;9)(q13;q34)	A	See Chromosome 9, Translocation
Chromosome 13									
503	2	A	66	1	M		46,XY/47,XY,+13	B	70%/30%
Chromosome 18									
143	3	A	107	3 mo.	F	B	47,XX,+18	A	Autopsy specimen
734	3	A	95	F	F		47,XX,+18	A	
1359	3	A	107	1 mo.	M	W	47,XY,+18		
Chromosome 21									
2504	7	A	95	1 mo.	M	B	47,XY,+21	A	Formerly GM-258
201*	10	A	30	13	F	W	47,XX,+21,t(1;17)(p32;p13)	A	See Translocation, Chromosome 1
144*	5	A	12	34	F	W	46,XX,der(6)t(6;21)(p25;q11)mat	A	See Translocation, Chromosome 6 and 21; Down's Syndrome

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 10; 6

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
							TRISOMY/POLYSOMY		
Chromosome 21, continued									
260	10	A	95	11	M	W	46,XY/47,XY,+21		Mosaic; 25% +21
1918	6	B	61	28	M	B	47,XY,+21,inv(9)(p13q21)	B	See Inversion Chromosome 9
1920	2	B	61	23	M	W	47,XY,+21,inv(9)(p13q13)mat	B	See Inversion Chromosome 9
2067	4	H	95	5 mo.F	M	W	47,XY,+21	A	
2058	5	B	165	11 da.	F	W	46,XX/46,XX,t(21;21)	B	70%/30% in leukocytes; see Translocation Chromosome 21
Chromosome 22									
2325	4	B	182	11 da.	F	W	47,XX,+22q-		Son of GM-2324, see Lymphoid, Translocation 16
84	5	A	95	1 mo.	M	W	47,XY,+22		
Chromosome X									
254	8	A	75	10 wk.F	F	B	47,XXX	A	76% interphase nuclei have 2 sex chromosome bodies; 15% have one; no evidence for mosaicism
1415	3	A	61	27	F	W	48,XXXX	A	4% Polyploidy
326	3	A	125	6	M	W	49,XXXXY	A	Severe retardation; Hypogonadism
1534	10	C	2	26	F	W	49,XXXXX		

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
							TRISOMY/POLYSOMY		
Chromosome X, continued									
157*	11	A	30	28	M	W	49,XXXXY,t(4;11)(q35;q23)pat	A	See Translocation, Chromosome 4
324	5	A	125	22	M	W	47,XXY	A	Klinefelter's; homo for G6PD deficient, med. type
325	4	A	125	30	M	W	47,XXY		Klinefelter's; hetero for G6PD def., med. type
Chromosome Y									
1250	3	C	106	23	M	B	47,XYY	A	Father of GM-1253 see Inversion Chromosome 9
							TRIPLOIDY		
1322	3	A	95	2 da.	F	B	69,XXX		Karyotype based on leukocytes; biopsy taken at autopsy
805	6	A	2	15 wk.F	M	I	69,XXY	B	Sib is 47,XY,+21; placental tissue
1672	8	J	88	1 da.	M	W	69,XXY	B	
							DELETION/RING/ISOCHROMOSOME		
Chromosome 1									
214	6	A	30	2	M	W	46,XY,del(1)(q42)	B	Growth hormone deficiency and hypothyroidism
803	3	C	2	2	M	W	46,XY,del(1)(q24q25)	B	Karyotype based on lymphocytes
2025	4	B	126	2 1/2	M	W	46,XY,del(1)(pter►q21or22::q25►qter)	A	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #23

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
							DELETION/RING/ISOCHROMOSOME		
Chromosome 2									
1138*	3	A	95	1	M	B	46,XY,del(2)(pter►p23)	A	
945	2	C	77	1 1/2	F	O	46,XX,del(2)(qter►p24:)	A	
Chromosome 3									
366	3	A	3	4	M	W	46,XY,rec(3),dup q inv(3)(qter►q21::p25►qter)	A	
Chromosome 4									
72	15	A	15	11	M	W	46,XY,del(4)(pter►p14:)	A	
343	3	A	66	3	M		46,XY,del(4)(p)		Wolf-Hirschhorn Syndrome
2003	4	B	95	11	F	W	46,XX,4q-		
Chromosome 5									
71*	15	A	15	19	F	W	46,XX,-5,der(5),t(5;?)(p1;?)mat	A	Deletion assumed to be derived rather then a simple deletion; see also Trans. Chromosome 5; ATCC CCL 90
Chromosome 6									
109	7	A	134	2	F	W	46,XX,r(6)	B	36% of cells have r(6)
Chromosome 8									
1549	8	A	140	7	M		46,XY,del(8)(p)	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 43; 45

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
					DELETION/RING/ISOCHROMOSOME				
Chromosome 9									
870*	2	C	15	22	F	W	46,XX,del(9)(qter➤p13:)	A	
166*	4	A	107	6	M	W	46,XY,r(9)(p2q3)	A	44% of cells have r(9); 46% are 45,XY,-9
1667	7	C	2	13	F	W	46,XX,del(9)(p22)	B	
1893	4	B	140	3	M		46,XY,del(9)(pter➤q13::q22➤qter)	A	
2356	2	B	140	3	M		46,XY,del(9)(pter➤p21:)	B	
Chromosome 11									
2008	6	B	69	2 wk.	F	B	46,XX,del(11)(q23)	B	
Chromosome 13									
509*	6	A	143	1 1/4	F	B	46,XX,del(13)(pter➤q14:)	A	Short term lymphs show r(13); formerly GM-250*
285	3	A	107	4 da.	M	B	46,XY,r(13)	A	
729	8	A	142	14	M	W	46,XY,r(13)	A	
Chromosome 16									
2346	2	B	204	3 mo.	M	W	46,XY,del(16)(q22)	B	
Chromosome 18									
1118	8	C	150	2 1/2	F	W	46,XX,r(18)	B	

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #'s 40; 21; 20

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
							DELETION/RING/ISOCHROMOSOME		
Chromosome 18, continued									
1727	13	A	142	45	M	W	46,XY,del(18)(p)		
Chromosome X									
735	17	A	77	17	F	W	46,Xqi,Xdic,inv(2)(p15 q21)	B	See Inversions, Chromosome 2
339	12	A	30	25	F	W	45,X/46,X,i(Xq)		
88*	4	A	12	19	F	W	46,X,i(X)(Xqter ► cen ► Xqter)	A	Buccal smear= 18% sex chromatin +; Turner Syndrome stigmata
1941	3	B	12	13	F	W	45,X/46,X,del(X)(q11)		In peripheral blood leukocytes
Chromosome Y									
1441	2	A	74	3 1/2	M	W	45,X/46,X,iso(Y)		
1709	4	C	109	11	F	W	46,X,i(Yq)	B	Gonadal dysgenesis
							TERATOMA		
1304	4 IMR	C	56	27	F		46,XX	B	PGM1=1;PGM3=1;PGD=AB; chromosome polymorphisms: 3=+/+;13=+/+;15=+/+;21=+/+; right ovarian teratoma; same patient
1305	4 IMR	C	56	27	F		46,XX	B	PGM1=1;PGM3=1;PGD=AB; chromosome polymorphisms: 3=+/-;13=+/-;15=+/-;21=+/-; uterine tissue same patient

*Publ'd. in Cytogenet. & Cell Genet.; see Appendix C ref. #5

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
							TERATOMA		
1306	8 IMR	C	56	31	F	W	46,XX	B	PGM1=1;PGM3=2-1;PGD=A; chromosome polymorphisms: 13=-/-;14=+/+;21=-/-; right ovarian teratoma; same patient
1307	5 IMR	C	56	31	F	W	46,XX	B	PGM1=1;PGM3=2-1;PGD=A; chromosome polymorphisms; 13=+/-;14=+/-;21=+/-; fallopian tube tissue; same patient
							GONADAL DYSGENESIS		
48	7	A	95	1	F	W	46,XY		Right gonadal tissue
83	7	A	95	1	F	W	46,XY	B	Left gonadal tissue — Same patient
868	2	C	107	26	F	W	46,XY		Gonadal dysgenesis; type unknown
978	7	C	143	15	F	W	46,XY/45,XO		Gonadal tissue
1491	3	A	95	8	F	W	46,XY		Phenotypic female
1709	4	C	109	11	F	W	46,X,i(Yq)	B	Phenotypic female
							BIOCHEMICAL MARKERS		
1150	4	C	125	23	M	W		A	Gd(+) ;Xg(a-); Sib
1152	4	C	125	46	M	W		A	Gd(-) ;Xg(a+); Father
1163	4	C	125	15	M	W		A	Gd(-) ;Xg(a+); sib of GM-1150; son of GM-1152

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
						BIOCHEMICAL MARKERS			
1153	4	C	125	42	M	W		A	Gd(+) ;Xg(a+); Father
1154	4	C	125	37	F	W		A	Phase=Gd Xg/Gd Xg ; Mother
1151	3	C	125	12	M	W		A	Gd(-) ;Xg(a-); Proband
1165	2	C	125	36	F	W			Phase=Gd Xg/Gd Xg
1166	3	C	125	39	F	W			Phase=Gd Xg/Gd Xg
1188	3	A	125	53	M	W	46,XY		Gd ; Xg(a+); karyotype based on leukocytes; Proband
1157	5	C	125	42	F	W	46,XX		Gd(+) Xg/Gd Xg(a+); Mother
1192	7	A	125	70	F	W	46,XX	A	Gd(+) /Gd(-) ;Xg(a+); Mother; karyotype based on blood; 100% G6PD(+) in red blood cells and 2 cell lines in hair roots
1193	4	A	125	50	F	W	46,XX	A	Gd(+) /Gd(-) ;Xg(a+); Proband; karyotype based on blood; 100% G6PD(-) in red blood cells, and 2 cell lines in hair roots
1717	3	B	125	39	M	W	46,XY		Xg(a-)
1862	2	B	125	37	M	W	46,XX		Xg(a-); Proband; Phenotypically male
1863	3	B	125	46	M	W	46,XY		Xg(?); Brother
1864	3	B	125	11	M	W	46,XY		Xg(-); Brother
1865	3	B	125	36	M	W	46,XX		Xg(+); Phenotypically male

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Paris Nomenclature	Verified	Remarks
							BIOCHEMICAL MARKERS		
1869	2	B	125	15	M	W			Xg(-)Gd(-)
1870	2	B	125	12	M	W			Gd(+)Xg(-)
1973	4	B	125	67	F	W	47,XXX		Xg(a+),G6PD mutant; see Trisomy X
1871	3	B	125	47	F	W			Double heterozygote in repulsion; Gd(B)Xg(-)/Gd(Med) Xg(+)
324	5	A	125	22	M	W	47,XXY		Klinefelter's; homo G6PD def.; Med. type
325	4	A	125	30	M	W	47,XXY		Klinefelter's hetero G6PD def.; Med. type
543	12	A	75	7	M	P	46,XY,1qh+	B	Duffy blood group; mental retardation; heterochromatic marker

APPARENTLY NORMAL HUMAN FIBROBLAST CELL CULTURES

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Verified*	Remarks
				NON-FETAL TISSUE**				
970	4	C	95	3 da.	M	W		
41	5	A	45	3 mo.	F	W	A	46,XX
302	6	A	95	10 mo.	M			
969	4	C	95	2	F	W	B	46,XX
498	4	A	128	3	M			
497	8	C	128	4	M	W		
408	8	A	128	5	F	W	A	46,XX
409	8	C	128	7	M	W	A	46,XY
499	7	A	128	8	M	W		
38	5	A	45	9	F	B	A	46,XX
500	8	A	128	10	M	W		
1652	7	A	133	11	F	W		
2036	5	J	133	11	F	W		
323	4	A	125	11	M	W		Brother of GM-326, (XXXXY)
316	3	A	26	12	M	W		See Aging Repository
1651	7	C	133	13	F	W		
2037	8	J	133	13	M	W		
37	4	A	45	18	F	W	A	
726	3	A	26	26	F	O		
975	3	C	26	26	F	W		
495	5	A	45	29	M		A	46,XY
23	5	A	95	31	F	W	A	46,XX; mother of Down's child
24	5	A	95	31	M	W	A	46,XY; father of Down's child
43	5	A	45	32	F	B	A	46,XX
185	6	A	76	33	F	W	A	46,XX; family history is positive for increased cholesterol
964	2	C	26	33	M	W	A	46,XY

* Verification indicates normal human karyotype by banding
**See also Family Groups listing

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
					NON-FETAL TISSUE			
2185	6	B	186	36	M	W		Normal spouse of a Huntington's Chorea patient
1650	6	A	133	37	F	W		
1653	8	A	133	37	M	W		
321	3	A	125	40	F	W	A	46,XX; Mother of GM-326 (XXXXY) and GM-323
322	4	A	125	40	M	W		Father of GM-326 (XXXXY)
275	3	A	26	42	M	W	A	46,XY; see Aging Repository
730	3	A	26	45	F	W	A	46,XX; see Aging Repository
288	4	A	35	64	M	W		
1681	8	A	133	70	M	W		
1680	8	A	133	71	F	W		
1706	5	A	26	82	F	W	A	46,XX; see Aging Repository
731	3	A	135	94	M	W		See Aging Repository
					Family Groups**			
41	5	A	45	3 mo.	F	W	A	46,XX; Daughter
37	4	A	45	18	F	W	A	Mother; see GM-637; SV40 trans.
43	5	A	45	32	F	B	A	46,XX; Mother
38	5	A	45	9	F	B	A	46,XX; Daughter
323	4	A	125	11	M	W		Son; Brother of GM-326 (XXXXY)
321	3	A	125	40	F	W	A	46,XX; Mother of GM-326 (XXXXY)
322	4	A	125	40	M	W		Father of GM-326 (XXXXY)
1652	7	A	133	11	F	W		Daughter
1651	7	C	133	13	F	W		Daughter
1650	6	A	133	37	F	W		Mother
1653	8	A	133	37	M	W		Father
1681	8	A	133	70	M	W		Paternal grandfather
1680	8	A	133	71	F	W		Paternal grandmother

**See also preceding page

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
				FETAL TISSUE (SEE ALSO HUMAN AMNIOTIC FLUID CELL CULTURES)				
9	3	A	26	3 mo.	M		A	46,XY; see Aging Repository
10	2	A	26	3 mo.	M	W	A	See Aging Repository
								46,XY, skin — Same
1380	8	C	26	3 mo.	M	W	A	Lung, 12cm crown-rump, 46,XY — fetus
11	2	A	26	2 mo.	M		A	See Aging Repository
468	2	A	26		M			14.5 cm, crown-rump
1379	2	C	26	3 mo.	M			Lung, 10 1/2cm crown-rump — Same
1381	4	C	26	3 mo.	M			See Aging Repository — fetus
1603	2	A	26	3 mo.	M	B		Skin — Same
1604	2	A	26	3 mo.	M	B		Lung; see Aging Repository — fetus

HUMAN AMNIOTIC FLUID CELL CULTURES

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				BIOCHEMICAL MUTANT CONDITIONS					
Cystinosis - 21980									
804	3	C	121	22 wk.F	M	W	--	B	46,XY
Fabry Disease (Diffuse Angiokeratoma) - 30150									
636	8	T	17	18 wk.F	M	W	y-	B	46,XY
Galactosemia (Transferase Deficiency) - 23040									
1743	3	B	77	6 mo.F	M	W	--		See GM-1741 Fibroblast
Homocystinuria (Cystathionine Synthase Deficiency) - 23620									
919	6	C	38	30 wk.F	F		--		46,XX
Hunter Syndrome - 30990									
1584	6	B	77	5 mo.F	M	W	y-	A	46,XY
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800									
236	5	B	85	16 wk.F	M		y-	B	46,XY
2338	2 IMR	C	183		M		y-		See GM-2290 & 2291 Fibroblast & GM-2292 Lymphoid
Metachromatic Leukodystrophy - Infantile - 25010									
2095	3 IMR	B	105	18 wk.F	F	W	--		American Indian
2214	4 IMR	B	105	20 wk.F	M	W	--		Probable
				CHROMOSOMAL ABERRATIONS					
Translocation									
477	9	C	48	14 wk.F	F	W			t(13;18) with trisomy 13q

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
				CHROMOSOMAL ABERRATIONS				
Inversions								
2029	5 IMR	A	172	13 wk.F	F			46,XX,inv(9)
2055	7	A	172	17 1/2 wk.F	F			46,XX,inv(9)
Trisomy								
1993	7	A	172	21 wk.F	M	W	A	47,XYY
2269	9	C	109	18 wk.F	M	W		47,XXY
				APPARENTLY NORMAL				
472	9	E	58	19 wk.F	M		B	46,XY
473	6	M	58	17 wk.F	M		B	46,XY
474	10	M	58	18 wk.F	M		A	46,XY
956	4	E	68	17 wk.F	F		B	46,XX
957	3	E	68	18 wk.F	F		B	46,XX
1420	11	C	109	18 wk.F	F	B	B	46,XX

HUMAN LYMPHOCYTE CULTURES WITH
BIOCHEMICAL MUTANT CONDITIONS

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
								DISORDERS OF AMINO ACID METABOLISM
								Immunoglobulins determined by Immunodiffusion.
Argininemia - 20780								
2011	E	6	5	F	W	--	B	IgG wk; Kappa+; Lambda wk; Iga+
Citrullinuria (Citrullinemia) - 21570								
235	E	10	31	M	W	--	B	
1204	E	107	8 mo.	M	W	--		Proband; See GM-1044 Fibroblast; (biochemically confirmed) IgG wk
1205	E	107	26	F	W	+-(O)		Mother; IgG+, Kappa wk
1206	E	107	27	M	W	+-(O)		Father
1207	E	107	4 1/2	F	W	+?		See GM-1058 Fibroblast; IgM wk Sib; IgG+, IgM+, Lambda+, Kappa wk
1685	E	157	1 da.	M		--		See GM-1679 Fibroblast
Cystathionuria - 21950								
1454	E	8	7		O	--	B	Unresponsive to B6
1456	E	8	43		W	+-	B	IgA+, Kappa+
1566	E	8	16		W	--	B	
1562	E	8	42		W	+-	B	IgA wk, IgM wk
1781	E	8	18		W	--	B	Proband; IgG+, IgM wk, Lambda wk; B6 responsive
1461	E	8	39		W	+-	B	Parent; IgA+, IgG+, Kappa+, IgM wk
1807	E	8	Adult	M	W	+-	B	Father
1565	E	8	16	F	W	--	B	Proband, also has PKU, IgW wk, IgM+, IgG wk

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF AMINO ACID METABOLISM				
Cystathionuria, continued								
1868	E	8	49		W	+-(O)		
Homocystinuria (Cystathionine Synthase Deficiency) - 23620								
1446	E	8		F	W	+-	B	IgA+, Lambda wk
1447	E	8		F	W	+-	B	Kappa+
1463	E	8	42	F	W	+-	B	Kappa+
1558	E	8		F		+-(O)		Kappa+, IgM+
1559	E	8		M		+-(O)		IgG+, Kappa wk
1560	E	8		M		--	B	Kappa wk
1528	E	8		M		+-(O)		IgA wk
1529	E	8	43	F		+-(O)		IgA+, IgG wk
1532	E	8		M		--		Kappa wk, IgM+
1808	E	8	45	M	W	+-	B	IgG wk, Kappa+
Maple Syrup Urine Disease - (Branched-Chain Ketoaciduria) - 24860								
1366	E	107	7	F	B	--		See GM-1364 Fibroblast; IgG+, Lambda wk
1655	E	107	5	F	W	--		See GM-1654 Fibroblast; IgG+
Phenylketonuria - 26160								
1565	E	8	16	F	W	--		Proband, also has Cystathionuria
1807	E	8	Adult	M	W	+-(O)		Father
				DISORDERS OF CARBOHYDRATE METABOLISM				
Fucosidosis - 23000								
1023	E	8	31	M	W	+-	B	Father IgA+, Kappa+
1024	E	8	9	M	W	--	B	Sib; Proband IgA+, Kappa+
1025	E	8	4	M	W	--	B	Sib; Proband IgA+, Kappa wk
1026	E	8	29	F	W	+-	B	Mother; IgG+

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
					DISORDERS OF CARBOHYDRATE METABOLISM			
Galactosemia (Transferase Deficiency) - 23040								
148	E	10	8	M	W	--	B	Proband; IgG+, Lambda+
149	E	10	47	M	W	+-	B	Father; IgG wk
2412	E	8	15	M	W	--		Proband; formerly GM-1027
1028	E	8	16	F	W	--		Sib
1029	E	8	41	F	W	+-(O)		Mother; IgG wk
Glycogen Storage Disease								
Type II - Pompe Disease - 23230								
1778	E	8	16	F	W	+-	B	Daughter; Kappa+, IgM+
1464	E	8	46	F	W	--	B	Proband
1568	E	8	71	M	W	+-	B	Father
Mucopolysaccharidosis								
Type IH - Hurler Syndrome - 25280								
1034	E	8	2 1/2	F	W	--	B	
1867	E	8	5	F	W	--	B	
Type IH/S - Hurler/Scheie								
1032	E	8	15	M	I	--	B	IgA+
Type IIIA - Sanfilippo A - 25290								
1780	E	8	8	M	W	--	B	Kappa wk, IgM+
Type VI - Maroteaux-Lamy Syndrome - 25320								
1022	E	8	4	F	W	--	A	IgA+, see GM-519 Fibroblast
					DISORDERS OF LIPID METABOLISM			
Abetalipoproteinemia - 20010								
1453	E	8	18	F	W			IgG wk, Kappa+
1810	E	8	23	M	W			

DISORDERS OF LIPID METABOLISM

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
Gaucher Disease								
Types 1 & 3 (Juvenile & Adult, Cerebral) - 23100								
1019	E	8	35	M	W	+-	B	Father of a Juvenile Gaucher; IgG+, Lambda+
1020	E	8	49	F	W	+-	B	Mother of an Adult Gaucher; IgG+, Kappa+
1021	E	8	25	M	W	--	B	Adult Gaucher; IgA+, Kappa wk
1030	E	8	50	F	W	+-	B	Mother of a Juvenile Gaucher; IgA+, Kappa+
1031	E	8	53	M	W	+-	B	Father of a Juvenile Gaucher
Hyperlipidemia - 14425								
1455	E	8	33	M		+-	B	IgG wk, Kappa+
1783	E	8	31	F		+-(0)		Combined
Hyperlipoproteinemia - 14440								
Type II - Familial Hypercholesterolemia								
1448	E	8	18	F	W	+-(0)		IgA+
1766	E	8	23	F	W	+-(0)		IgA+, Kappa wk, formerly GM-1449
1450	E	8	49	M	W	+-(0)		
1767	E	8	19	F		+-(0)		Formerly GM-1452
1458	E	8	30	F		--		Kappa wk
1459	E	8		F		--		IgG+, Lambda wk
1460	E	8		F		+-(0)		IgG wk, Kappa+
1567	E	8	28	M	B	--		Kappa wk
1784	E	8	23	M		--		
Metachromatic Leukodystrophy - 25000								
1017	E	8	14	F	W	--	B	Kappa wk, IgM+
1018	E	8	25	F	P	+-	B	IgA+, Kappa+
1016	E	8	42	M	W	+-(0)		Father of a Juvenile MLD
1785	E	8	14	F	W	--		Juvenile

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF NUCLEOTIDE AND NUCLEIC ACID METABOLISM				
HGPRT Deficiency								
467	E	83		F				Induced with ethylmethane sulfonate, 6-thioguanine resistant
Inosine Triphosphate Pyrophosphohydrolase (ITPase) Deficiency								
1619	E	177	29	F	W			Kappa+, IgM+; See GM-1617 Fibroblast
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800								
1899	E	66	10	M	W	y-		Proband
1900	E	66	Adult	F	W	+-(0)		Mother; Kappa+
2292	E	183	F	M		y-		See GM-2290-2291 Fibroblasts; see GM-2338 Amniotic; fetal tissue
Xeroderma Pigmentosum - 27870								
1646	E	133	21	F	W	--		See GM-1389 Fibroblast, Kappa+, IgM+
				OTHER DISORDERS OF KNOWN BIOCHEMISTRY				
Menkes Syndrome (Kinky Hair Disease) - 30940								
1245	E	107	5 da.	M	W	y-		Sib of GM-220; See GM-1057 Fibroblast; Kappa+, IgM+
1982	E	107	2	M	W	y-		Proband; see GM-1981 Fibroblast; Kappa wk, IgM+
1984	E	107	26	F	W	+-(0)		Mother; see GM-1983 Fibroblast; IgG+, Kappa+, IgM wk
Porphyria								
Acute Intermittent Porphyria - 17600								
1363	E	112		F	W	+-	A	
2133	E	112	65	M	W	+-	A	Father, see GM-941 Fibroblast, Kappa+, IgM+
2134	E	112	63	F	W	++	A	Mother, see GM-940 Fibroblast, IgG+, Kappa+
2135	E	112	25	F	W	+-	A	Proband; see GM-939 Fibroblast

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
						OTHER DISORDERS OF KNOWN BIOCHEMISTRY		
Porphyria								
Acute Intermittent Porphyria, continued								
2229	E	112	23	F	W	+-	B	
2230	E	112	58	F	W	+-	B	
						DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY		
Adrenal Hyperplasia (Adrenogenital Syndrome) Type III - 20191								
2242	E	107	9 mos.	F	W	--		See GM-2241 Fibroblast
Basal Cell Nevus Syndrome - 10940								
1553	E	104	27	M	W	+-(0)		IgA+, IgG+, Kappa+, Lambda wk
1576	E	104	31	F	W	+-(0)		IgG+, Lambda wk
1578	E	104	39	M	W	+-(0)		IgA wk, Lambda+, IgM+
1656	E	104	53	M	W	+-(0)		See GM-1657; GM-1658 Fibroblast; IgA wk, Kappa+, IgM+
1726	E	104	58	F	W	+-(0)		See GM-1725, Fibroblast; IgA+, IgG wk;
2139	E	104		F	W	+-(0)		See GM-2138 Fibroblast
2099	E	104	31	M	W	+-(0)		See GM-2098 Fibroblast; IgA+
Cockayne Syndrome - 21640								
1712	E	6	21	M	W	--		
Cystic Fibrosis (Mucoviscidosis) - 21970								
504	E	25	19	M	W	--	B*	Proband; Lambda+, IgM+
505	E	25	Adult		W	+-	B*	Parent; IgG wk, Kappa+
506	E	25	8	F	W	--	B*	IgA+
608	E	25	21	M	W	--	B*	IgG+, Kappa+
897	M	89	12 1/2	M	W	--		IgG+, Lambda wk

*Production of ciliary dyskinesis factor

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
					DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY			
Cystic Fibrosis, continued								
1442	E	8	31	M	W	+-	B*	Father
1443	E	8	28	F	W	+-	B*	Mother; IgA+, Lambda+
1445	E	8		F	W	--	B*	Proband; Kappa+
1530	E	8	5	M		--		Lambda+, IgM+
1531	E	8	23	M	W	--		IgG+, Kappa wk
1444	E	8	32	F		+-	B*	IgG+, Kappa wk
Diabetes Mellitus - 22210								
1241	E	34	37	M	W			See GM-1486 Fibroblast; maturity onset diabetes; IgG+
1817	E	137	65	M	W			MODY diabetes; primary affective disorder, biopolar; Lamda wk, IgM+
1905	E	34	32	F	W			MODY diabetes; Kappa+, IgM+
Family #1, Maturity Onset Diabetes - 22210								
1240	E	34	33	F	W			Sib; see GM-1122 Fibroblast; IgA+, Kappa+, Lambda+, IgM+
1246	E	34	46	M	W			Sib; see GM-1219 Fibroblast; IgG+, Kappa+, IgM+
1247	E	34	30	M	W			Sib; see GM-1237 Fibroblast; IgG+, Kappa+, IgM wk
1243	E	34	35	M	W			Sib; see GM-1430 Fibroblast; IgG+, Kappa+
1956	E	34	44	M	W			Sib; see GM-1955 Fibroblast, Kappa+, IgM+
1498	E	34		F	W			Sib; see GM-1497 Fibroblast; IgA wk, IgG+, Kappa+. IgM wk
1242	E	34	22	M	W			See GM-1435 Fibroblast; son of GM-1498; Lambda+, IgM+
1244	E	34	20	M	W			See GM-1496 Fibroblast; son of GM-1498; IgG+, Kappa+

*Production of ciliary dyskinesis factor

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
						DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY		
Diabetes Mellitus								
Family #1, Maturity Onset Diabetes, continued								
1410	E	34	15	F	W			See GM-1409 Fibroblast; mat. cousin of GM-1242-1244; only juvenile diabetic in family
1838	E	34	22	F	W			IgG+, Kappa+; pat second cousin of the 6 sibs; MODY type II; see GM-1837 Fibroblast
Family #2, Juvenile with Optic Atrophy - 22230								
1795	E	189	13	F	W			Optic atrophy; sister; see GM-1610 Fibroblast; Kappa+, IgM+
1796	E	189	16	M	W			Normal brother; IgG wk, Kappa+
1797	E	189	42	M	W			Normal father; see GM-1701 Fibroblast; Kappa+, IgM+
1799	E	189	18	F	W			Optic atrophy sister; see GM-1609 Fibroblast; Kappa+, IgM+
1800	E	189	6 1/2	F	W			Healthy sister, twin; IgG+
1801	E	189	6 1/2	F	W			Healthy sister, twin; Kappa+, IgM+, Lambda wk
1802	E	189	39	F	W			Normal mother; Kappa wk, Lambda+, IgM+
1803	E	189	11	F	W			Normal sister; Lambda+, IgM+
Dysautonomia (Riley-Day Syndrome) - 22390								
599	E	24		F	W	--		IgA+, Kappa+
600	E	24		M	W	+-(O)		IgM wk
601	E	24	10	M	W	--		Kappa+, IgM+
602	E	24		M	W	+-(O)		Kappa+, IgM+
603	E	24		F	W	+-(O)		IgG+
1465	E	8	21	F	W	--		IgM wk

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY				
Dysautonomia (Riley-Day Syndrome), continued								
1466	E	8	11	M	W	--		Kappa+, IgM+
1777	E	8	22	F	W	--		Kappa+, IgM+
Dyskeratosis Congenita - 30500								
1775	E	107	6 1/2	M	W	y-		See GM-1774 Fibroblast; Kappa wk
Dystonia Musculorum Deformans - 12810; 22450								
2217	E	184	30	M	W	+-(0)		Age of onset 24 yrs.; IgA+, IgG+, Kappa+, Lambda wk; See GM-2215 Fibroblast
2256	E	184	14	F		--		Recessive form, age of onset 12 yrs.; See GM-2255 Fibroblast
2264	E	184	14 1/2	F	W	+-(0)		IgG+; Jewish background, age of onset 8 yrs.
2305	E	184	16	F	W	+-(0)		Age of onset 7 yrs.; see GM-2304 Fibroblast
2307	E	184	13	M	W	+-(0)		Age of onset 10 yrs.; Jewish background, see GM-2306 Fibroblast
2347	E	184	21	F	W	+-(0)		Swedish background, age of onset 5 yrs.
Huntington Chorea - 14310								
2078	E	136	25	F	W	+?		At risk; daughter; see GM-2077 Fibroblast; IgA+
2080	E	136	48	F	W	+-		Proband; see GM-2079 Fibroblast; IgG+, Kappa wk
2146	E	186	54	M	W	+-		Proband; see GM-2147 Fibroblast
2148	E	186	54	F	W	++		Normal wife; see GM-2149 Fibroblast
2150	E	186	28	F	W	+?		Daughter; see GM-2151 Fibroblast

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
						DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY		
Huntington Chorea, continued								
2152	E	186	40	F	W	++		Normal widow of deceased proband; see GM-2153 Fibroblast
2154	E	186	20	F	W	+?		Daughter; see GM-2155 Fibroblast
2156	E	186	16	F	W	+?		Daughter; see GM-2157 Fibroblast
2158	E	186	19	F	W	+?		Daughter; see GM-2159 Fibroblast
2160	E	186	21	M	W	+?		Son; see GM-2161 Fibroblast
2162	E	186	12	M	W	+?		Son; see GM-2163 Fibroblast
2166	E	186	58	F	W	+-(O)		Affected sister of GM-2165, Fibroblast Kappa+, IgG+
2168	E	186	52	M	W			Normal spouse of GM-2166, see GM-2169, Fibroblast
2178	E	186	38	F	W	+?		Maternal niece of GM-2165,2166, daughter of GM-2186
2180	E	186	52	F	W	++		Normal spouse of GM-2165
2182	E	186	21	F	W	+?		Daughter of GM-2165, see GM-2183 Fibroblast
2186	E	186	60	F	W	+?		Sister of GM-2165, 2166; IgA+, Kappa+,Lambda+; see GM-2187 Fibroblast
2176	E	186	26	M	W	+?		Son of GM-2165, see GM-2177 Fibroblast
2188	E	186	63	M	W	++		Normal spouse of GM-2186, see GM-2189 Fibroblast
2170	E	186	22	F	W	+?		Daughter; see GM-2171 Fibroblast
2172	E	186	52	F	W	+-(O)		Proband; see GM-2173 Fibroblast
2174	E	186	55	M	W	++		Spouse; see GM-2175 Fibroblast
Hyper IgM								
390	E	10	3	F	W			

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verfied	Remarks
								DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY
Neurofibromatosis (Von Recklinghausen Disease) - 16220								
1634	E	104	61	M	W	+-(0)		Kappa wk, IgM wk; see GM-1633 Fibroblast; Atypical
1861	E	104	41	M	W	+-(0)		Kappa+, IgM+; see GM-1860 Fibroblast
1641	E	104	19	F	B	+-(0)		See GM-1639 Fibroblast; IgG+, Kappa+, IgM+
Persistance of Fetal Hemoglobin (Hemoglobin F) - 14170								
2064	E	192	53	M	B	+-(0)		Heterozygous for Elliptocytosis; Kappa+, IgM+
Schizophrenia and Psychiatric Disorders - 18150								
1487	E	137	53	F	W			Normal Mother
1488	E	137	23	M	W			Proband
1489	E	137	55	M	W			Schizophrenic; Father
1793	E	137	26	M	W			Affected son; see GM-1792 Fibroblast
1834	E	137	56	M	W			Proband; see GM-1833; Atypical Psychosis Fibroblast; Lambda+, IgM+
1836	E	137	27	F	W			Affected daughter; see GM-1835 Fibroblast; IgA+, IgG+, IgM+, Kappa+
1883	E	137	25	F	W			Normal daughter; see GM-1882 Fibroblast; IgG+, Kappa+, Lambda+, IgM+

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
				DISORDERS OF UNCERTAIN BIOCHEMICAL ETIOLOGY				
Schizophrenia and Psychiatric Disorders, continued								
1825	E	137	56	M	W			"Carrier" for schizo., father; see GM-1824 Fibroblast; IgA+, Kappa+, IgM+, Lambda wk
1827	E	137	24	F	W			Proband; Schizophrenia IgA wk, Kappa+, IgM+, IgG+
1847	E	137	20	M	W			Normal first cousin; see GM-1846 Fibroblast; son of GM-1845; IgG wk, Kappa +
1885	E	137	31	M	W			Normal brother; Lambda+, IgM+
1845	E	137	55	M	W			Affected uncle, father of GM-1847; See GM-1844 Fibroblast
Sea-Blue Histiocyte Disease - 26960								
1913	E	163	24	F	W	--		See GM-1912 Fibroblast; IgG+, Kappa wk
Tuberous Sclerosis - 19110								
1636	E	104	17	M	B	+-(0)		See GM-1635 Fibroblast; IgA wk, Kappa wk
1638	E	104	20	F	B	+-(0)		Kappa+, IgM+; see GM-1644 Fibroblast
Waldenstrom Macroglobulinemia - 15360								
1501	E	10	69	M		+-(0)		

HUMAN LYMPHOCYTE CULTURES WITH
CHROMOSOMAL ABERRATIONS

GM #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
						SYNDROME WITH INCREASED CHROMOSOME BREAKAGE		
Ataxia-Telangiectasia - 20890								
717	E	66	11	M	W	--		
718	E	66	9	M	W	--		Sib; Kappa+
719	E	66	13	F	W	--		Proband; Lambda+
736	E	66	Adult	M	W	+-(O)		Father; Kappa+
781	E	66	4	F	W	--		Sib; Kappa wk
1525	E	54	19	F	W	--		Sib; Lambda+, IgM+; 46,XX,t(14;14) clone in short term lymphocytes
1526	E	54	28	F	W	--		Proband; IgG wk, Kappa wk, IgM+; 46,XX,t(X;14) clone in short term lymphocytes
						TRANSLOCATIONS		
1203	E	66	30	F	W			46,XX,t(4;12); IgG wk, Kappa+, IgM+
1063	E	67	12 1/2	F	B			46,XX,t(4;13)(q21;q14); IgG wk
633	E	89	8	M	W			46,XY,t(7;10)(q2;q11);Kappa+, IgM+
1388	E	61	14	F	W			46,XX,t(9;13)(q22;q12)mat; IgA+, Kappa+
1561	E	8	36	F	W			46,XX,t(4;11)(q25;q13);IgA+, IgM wk
1261	E	107	13	F	W			45,XX,-13,-18,+t(13p;18p); IgA+, Kappa wk, Lambda+
2324	E	182	27	F	W			46,XX,t(16;22)(p13;q22); Mother of GM-2325 Fibroblast, see Trisomy 22; Kappa+
						TRISOMY/POLYSOMY		
1416	E	61	27	F	W			48,XXXX; IgM wk

GM #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
						TRISOMY/POLYSOMY	
1202	E	66	7	M	W		49XXXXY; Kappa+, IgM+
1201	E	144	1	F	W		45,XX,-21,+t(21;21) derived from a Down's Syndrome patient with 46,XX,t(21;21)
1919	E	61	28	M	B		47,XY,+21,inv(9)(p13q21); see GM-1918 Fibroblast
1921	E	61	23	M	W		46,XY,+21,inv(9)(p13q13)mat; see GM-1920 Fibroblast; Kappa+, IgM+

UNCLASSIFIED HUMAN LYMPHOCYTE CULTURES

GM #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
Allergic Asthmatic							
604	E	25	10	M	W		Kappa+
Leukemia							
463	E	10	60	M	W		Chronic Lymphocytic Leukemia
Multiple Myeloma							
1311	E	51	70	M	B		Bone marrow culture
1312	E	51	70	M	B		Kappa wk; Same patient
1500	E	10		M			IgG myeloma; IgG+, Lambda+

APPARENTLY NORMAL HUMAN LYMPHOCYTE CULTURES

GM #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
892	E	24	12	F	W		IgM+
1953	E	89	22	F	W		
946	E	24	22	F	W		Kappa+, IgM+
1079	E	89	22	F	W		
131	E	10	23	F	W		IgG+, Kappa+
333	E	10	23	F	W		IgG+, Kappa+
546	E	89	23	F	W		
607	E	25	23	F	W		Kappa+, IgM+
922	E	24	23	F	W		IgG wk, IgM+, Kappa+
924	E	24	23	F	W		
923	E	24	24	M	W		IgA+, Lambda+
605	E	25	24	F	W		IgG+, Kappa+
130	E	10	25	M	W		IgG+, Kappa+
558	E	89	26	M	W		IgM+
1805	E	89	26	F	W		
536	E	89	27	M	W		
621	E	89	28	M	W		IgG+
1078	E	89	30	F	W		IgG+
893	E	24	32	F	W		Lambda+
894	E	24	32	F	W		
1806	E	89	33	M	W		Formerly GM-1074
1989	E	89	33	M	W		Formerly GM-1076
2184	E	186	36	M	W		Normal spouse of HD patient, see GM-2185 Fibroblast
1814	E	89	37	F	W		Formerly GM-1072
1815	E	89	42	M	W		Formerly GM-1075
1954	E	89	44	F	W		Formerly GM-1080
1990	E	89	51	F	W		Formerly GM-1077
1310	E	89	51	M	W		IgA wk, IgG+, Lambda+, IgM wk
1056	E	26	65	M	W		IgA+, Lambda+
606	E	24	Adult		W		Kappa+
891	E	24	Adult				

SV40 VIRUS TRANSFORMED CELL CULTURES

GM #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
Galactosemia (Transferase Deficiency) - 23040									
638	48	C	51	3 mo.	M		--	A	SV40 transformed GM-54 culture, Gal-1-P uridyl transferase not present after crisis; G6PD-B, T-antigen positive; see GM-54 Fibroblast
639		C	51	8	M	B	--	A	SV40 transformed GM-52 culture, Gal-1-P uridyl transferase not present after crisis; G6PD-A; See GM-52 Fibroblast
Lesch-Nyhan Syndrome (HGPRT Deficiency) - 30800									
847		B	51	5 1/2	M	B	y-	A	SV40 transformed GM-177 culture; deficient for HGPRT, G6PD type A; see GM-2063 Fibroblast
Apparently Normal									
637	40	C	51	18	F	W		A	SV40 transformed GM-37 normal culture; T-antigen positive; See GM-37 Fibroblast

GM #	Passage #	Culture Media	Submitter Code	Verified	Remarks
Mouse					
86		K	40	A	Clone #745, Friend DMSO hemoglobin inducible secreting cell line
979		Q	158	A	Murine erythroleukemic line derived from a splenic focus in DDD mouse infected with Friend Leukemia virus; aneuploid and hypotetraploid; inducible for erythroid differentiation by DMSO
346		J	83	A	A9 cell line; deficient in HGPRT and selectable with 8-azaguanine
347		J	83	A	B82 cell line; deficient in thymidine kinase and selectable with 5-bromodeoxyuridine
Syrian Baby Hamster Kidney					
345		J	83	A	TG-2 cell line; deficient in HGPRT and selectable with 6-thioguanine
348		J	83	A	B1 cell line; deficient in thymidine kinase and selectable with 5-bromodeoxyuridine
511		J	83	A	A5 cell line; deficient in thymidine kinase but with increased folate reductase

GM #	Passage #	Culture Media	Submitter Code	Verified	Remarks
Chinese Hamster					
215		R	22	A	V79 young adult male cell culture established by C. Ford and G. Yerganion and used in somatic cell genetics to isolate drug resistant mutants (lung tissue)
458		J	53	A	CHW normal male Chinese hamster cell culture established by C.C. Lin, U. of Calagary from skin taken from the ear
459		J	53	A	CHW-1102 cell culture derived from CHW normal culture after treatment with methylmethanesulphonate and selected with 8-azaguanine; culture is deficient in HGPRT

Repository #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Remarks
Alzheimer Disease of Brain - 10430								
GM 364	2	A	139	53	M	W	+-	See GM Repository
Bloom Syndrome (See GM Repository) - 21090								
GM 1620	22	C	44	8 mo.	F	W	--	
Cowden Disease - 15835								
AG 1964	3	B	195	64	M	W	+-	Affected
AG 1966	3	B	195	58	F	W	+-	Affected
AG 1965	3	B	195	60	F	W	++	Normal — Family group
AG 1967	3	B	195		M	W	++	Normal
AG 1968	3	B	195	49	F	W	++	Normal
AG 1969	2	B	195		F	W	++	Normal
Lowe Oculocerebrorenal Syndrome - 30900								
AG 1756	3	B	94	2 1/2	M		y-	See GM Repository
Mulibrey Nanism - 25325								
AG 2122	7	A	4	11	F		--	
Progeria (See GM Repository) - 26410								
GM 917	13	C	49	17	F		--	
AG 989	11	A	55	20	M			Atypical; cachectic dwarfism
AG 990	9	C	55		M			Atypical
AG 991	7	A	55	4	M		--	Classical
GM 1177	8	C	49	9	M		--	
GM 1178	18	C	49	34	M		--	
AG 1972	8	B	91	14	F	W	--	
AG 1710	5	B	197		M			Atypical; progeria-like

Repository #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Remarks
Thanatophoric Dwarfism - 27365								
AG 711	2	A	143	1 da.	M	W		Cartilage — Same patient
AG 712	2	A	143	1 da.	M	W		Spleen — Same patient
AG 713	2	A	143	1 da.	M	W		Lung — Same patient
AG 714	3	A	143	1 da.	M	W		Skin — Same patient
Translocation								
AG 1839	7	B	31	10	F	W		45,XX,t(7q;13q); left side Hemihypertrophy; right side
AG 1840	7	B	31	10	F	W		Same patient
Xeroderma Pigmentosum (See GM Repository) - 27870								
AG 1951	12	C	201	40	M	W	--	From USSR
AG 1952	12	C	201	7	M	W	--	From USSR; DeSanctis Cacchione type
					TUMOR PATIENTS			
Chronic Myelogenous Leukemia								
AG 1731	3	B	94	19	F			Irradiated — Same patient
AG 1732	3	B	94	19	F			Non-irradiated; lymphocytes show Phila. marker chromosome; patient had Wilm's tumor previously
Glomus Tumor								
AG 716	3	A	159					

Repository #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Remarks
Multiple Myeloma								
AG 1311	3	E	156	55	M	B		Bone marrow — Same patient
AG 1312	3	E	156	55	M	B		Lymphocyte — Same patient
AG 1360	3	A	156	55	M	B		Skin — Same patient
AG 1485	3	A	156	55	M	B		Skin — Same patient
Neuroblastoma								
AG 2202	3	B	94	9	F			Non-irradiated skin — Same patient
AG 2203	3	B	94	9	F			Irradiated skin — Same patient
AG 2272	3	B	94					
Osteogenic Sarcoma								
AG 2086	2	B	94	20	M			Skin
Retinoblastoma - 18020								
GM 913	3	C	65	2	M	W	++	See GM Repository; skin, does not grow well — Same patient
GM 914	3	C	65	2	M	W	++	Conjunctiva; sporadic — Same patient
AG 1142	4	C	94	2 1/4	F	W		46,XX,del(13)(q14q22) — Same patient
AG 1484	3	E	94	2 1/4	F	W		Lymphoid culture — Same patient
AG 1231	3	C	65	2	F	W	++	Conjunctiva; sporadic — Same patient
AG 1232	3	E	65	2	F	W	++	Tumor — Same patient
AG 1979	4	J	198	2	M		++	Conjunctiva; sporadic
AG 1946	6	J	198	1 1/2	M			Conjunctiva; sporadic
AG 1947	6	J	198	2	M			Conjunctiva; sporadic
AG 1123	3	A	94	9 mo.	F	B	+-	Conjunctiva; ident. twin hereditary
AG 1131	3	C	94	9 mo.	F	B	+-	Conjunctiva; ident. twin hereditary
AG 1223	8	C	161		F	B	+-	Conjunctiva; same patient as GM-1131
AG 1408	4	A	65	8 mo.	F	W	+-	Conjunctiva; hereditary
AG 1978	4	J	198	2	M		+-	Conjunctiva; hereditary
AG 1980	4	J	198	1	F		+-	Conjunctiva; hereditary
AG 1262	4	C	198	7 wk.	F	W	+-	Conjunctiva; hereditary

Repository #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Genetic Status	Verified	Remarks
Wilms Tumor									
AG 1615	3	B	160	9	M	B/W			Non-irradiated skin] Same
AG 1616	3	B	160	9	M	B/W			Irradiated skin] patient
AG 2089	6	B	94	19	F				
AG 2129	2	B	199	10	M	B			Fibroblast culture] Same
AG 2130		E	199	10	M	B			Lymphoid line] patient
AG 1894	2	B	94	12	M	B			Irradiated skin] Same
AG 1895	2	B	94	12	M	B			Non-irradiated skin] patient
APPARENTLY NORMAL CULTURES									
GM 11	2	A	26	2 mo.F	M			A	See GM Repository, 46,XY
GM 1379	4	C	26	3 mo.F	M	W		A	Lung, Puerto Rican, 46,XY] Same
GM 1381	4	C	26	3 mo.F	M	W		A	See GM Repository, 46,XY] fetus
GM 9	3	A	26	3 mo.F	M			A	See GM Repository, 46XY
GM 10	2	A	26	3 mo.F	M	W		A	See GM Repository, 46XY] Same
GM 1380	8	C	26	3 mo.F	M	W		A	Lung, 46XY] fetus
GM 1603	2	A	152	3 mo.F	M				See GM Repository;] Same
GM 1604	2	A	152	3 mo.F	M				Lung] fetus
AG 1437	2	A	152	3 da.	M	W			Foreskin
AG 1439	3	A	152	3 da.	M	B			Foreskin
AG 1440	4	A	152	3 da.	M	B			Foreskin
AG 1518	2	A	152	3 da.	M			A	Foreskin: 46,XY
AG 1519	2	A	152	3 da.	M			A	Foreskin: 46,XY
AG 1520	2	A	152	3 da.	M				Foreskin
AG 1521	2	A	152	3 da.	M				Foreskin
AG 1522	2	A	152	3 da.	M				Foreskin
AG 1523	2	A	152	3 da.	M			A	Foreskin; 46,XY
GM 316	3	A	26	12	M	W		A	See GM Repository
AG 2220	3	B	200	22	M	W			
GM 275	3	A	26	42	M	W		A	See GM Repository
GM 730	3	A	26	45	F	W		A	See GM Repository

Repository #	Passage #	Culture Media	Submitter Code	Age	Sex	Race	Verified	Remarks
				APPARENTLY NORMAL CULTURES				
AG 2257	1	B	200	46	F	W		Skin] Same
AG 2258	4	B	200	46	F	W		Lung] patient
AG 2222	2	B	200	49	M	W		Skin fibroblasts
GM 1706	4	A	26	82	F	W	A	See GM Repository; formerly GM-237
GM 731	3	A	26	95	M	W	A	See GM Repository
				SPECIALLY CHARACTERIZED DIPLOID CELL CULTURES*				
IMR 90	1	A	26	16 wk.F	F	W	A	Lung; see guidelines for distribution of these cells, Science 196:60-63 (1977)
IMR 91	1	A	26	14 wk.F	M	W	A	Lung] Same
IMR 91	1	A	26	14 wk.F	M	W	A	Skin] fetus

*Large quantity of characterized cells frozen at early passage

CODE FOR CULTURE MEDIA

For reference see: Morton, Helen C., A survey of Commercially Available Tissue Culture Media. In Vitro 6:89-108, 1970.

A - McCoy's 5A with 20% fetal bovine serum (FBS) not inactivated (Iwakata and Grace modification)

B - Ham F12 with 20% FBS not inactivated

C - Minimum Essential Medium Eagle in Earle's BSS with 20% FBS not inactivated (with 2x concentration of essential and non-essential amino acids and vitamins)

D - Roswell Park Memorial Institute 1640 with 10% FBS inactivated 60°C for 1/2 hour

E - Roswell Park Memorial Institute 1640 with 20% FBS inactivated 60°C for 1/2 hour

F - Minimum Essential Medium Eagle in Earle's BSS with 30% FBS not inactivated (with 2x concentration of essential and non-essential amino acids and vitamins)

G - Ham F12 with 10% FBS not inactivated

H - McCoy's 5A with 10% FBS not inactivated (Iwakata and Grace modification)

I - Roswell Park Memorial Institute 1640 with 20% FBS not inactivated

J - Minimum Essential Medium Eagle in Earle's BSS with 10% FBS not inactivated (with 2x concentration of esssential and non-essential amino acids and vitamins)

K - Minimum Essential Medium Eagle in Earle's BSS with 15% FBS not inactivated (with 2x concentration of essential and non-essential amino acids and vitamins)

L - Minimum Essential Medium Eagle in Earle's BSS with 12% FBS not inactivated (with 2x concentration of essential and non-essential amino acids and vitamins)

M - Roswell Park Memorial Institute 1640 with 30% FBS inactivated 60°C for 1/2 hour

N - Minimum Essential Medium Eagle in Earle's BSS with 10% FBS inactivated 60°C for 1/2 hour (with 2x concentration of essential and nonessential amino acids and vitamins)

O - Roswell Park Memorial Institute 1640 with 30% FBS not inactivated

P - Ham F12 with 10% FBS inactivated 60°C for 1/2 hour

Q - Ham F12 with 20% FBS inactivated 60°C for 1/2 hour

R - Minimum Essential Medium Eagle in Earle's BSS with 5% FBS not inactivated (with 2x concentration of essential and non-essential amino acids and vitamins)

S - Minimum Essential Medium in Hanks' BSS with 10% FBS not inactivated (with 2x concentration of essential and non-essential amino acids and vitamins)

T - Minimum Essential Medium in Hanks' BSS with 20% FBS not inactivated (with 2x concentration of essential and non-essential amino acids and vitamins)

U - Ham F12 with 30% FBS not inactivated

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41. A (5;14) translocation, 46 chromosomes. Repository identification No. GM-589. D.S. Borgaonkar, M.M. Aronson, A.E. Greene, L.L. Coriell. Cytogenet. Cell Genet. 18:242 (1977).

42. A (9;17) translocation, 46 chromosomes. Repository identification No. GM-988. C.C. Lin, A.J. Kavanagh, M.M. Aronson, A.E. Greene, L.L. Coriell. Cytogenet. Cell Genet. 18:244 (1977).

43. Deletion in the short arm of chromosome No. 2 from a subject with congenital anomalies. Repository identification No. GM-1138. E. Zackai, B. Emanuel, W.J. Mellman, M.M. Aronson, B. Bozarth, A.E. Greene, L.L. Coriell. Cytogenet. Cell Genet. 18:108 (1977).

44. A (2;8) translocation, 46 chromosomes. Repository identification No. GM-327. R. Worton, M.M. Aronson, A.E. Greene, L.L. Coriell. Cytogenet. Cell Genet. 18:243 (1977).

45. Deletion of the short arm of chromosome 4 from a subject with Wolf's syndrome. Repository identication No. GM-72. W.R. Breg, M.M. Aronson, A.E. Greene, L.L. Coriell. Cytogenet. Cell Genet. 18:307-308 (1977).

46. Citrullinemia, argininosuccinate synthetase deficiency. Repository identification No. GM-1044. E. Spector, N. Kennaway, H. Punnett, A.E. Greene, L.L. Coriell. In Press.

47. A (4;7) translocation, 46 chromosomes. Repository identification No. GM-773. L.G. Jackson, B. Bozarth, M.M. Aronson, A.E. Greene, L.L. Coriell. In Press.

48. Duarte variant-galactosemia heterozygote. Repository identication No. GM-1996. G.N. Donnell, W.G. Ng, O.S. Alfi, A.E. Greene, L.L. Coriell. In Press.

49. A (4;7) translocation, 46 chromosomes. Repository identification No. GM-1220. L.G. Jackson, B. Bozarth, M.M. Aronson, A.E. Greene, L.L. Coriell. In Press.

HL-A antigens of human lymphocyte cell cultures listed on pages 107 through 122. The Human Genetic Repository is indebted to Dr. Roger Kennett, Department of Human Genetics, University of Pennsylvania Medical School, Philadelphia, Pennsylvania.

For further data showing reactions with a selected set of NIH antisera, contact Dr. Kennett.

GM #	HL-A	GM #	HL-A
235	(Aw31,33)A9,Bw15,B12	1366	A2(A9),B8,B12
558	A3,A11,Bw15,Bw22	1410	A2,Aw24,Bw35,B13(B12)
892	A2(A9),B12,B18	1446	A2,3,Bw35,Bw17
1027	A2-B12/A1-B8	1447	A1,A10,B7,(Bw16)
1028	A1/A2-B12	1454	A28,A2,Bw40,Bw21
1029	A2-B12/A28-Bw35	1455	A11(A3),Bw22,(Bw40)
1032	A9,A10,B13	1456	A1,B13,Bw27
1056	A9,29,B12,Bw15	1461	A2,A3,Bw22,Bw40
1063	A1,B12,Bw17	1463	A2,A11,B27,Bw22
1204	A2-B12/A28-Bw35	1500	A2(A3),B12,Bw15
1205	A28-Bw35/A1-B13	1501	A3,A9,Bw22,(B12)
1206	A11-Bw15/A2-B12	1525	A28,(Aw31,33),B12
1207	A11-Bw15/A28-Bw35	1526	A9,A10,Bw17,B13(Bw40)
1241	A3,A10,B13,B12	1528	A10,(A3),B18,Bw22
1245	A9,A10,B13,B18	1529	A10,Aw23,B8,B18
1261	A1,A2,Bw15(B18)	1530	A2,A11,Bw17,Bw35
1311	A10,A11(A3),B12	1531	A2,(A9),B7,(Bw40)
1312	A10,A11(A3),B12	1532	A2,B7,Bw22,(B18)

GM #	HL-A
1539	A3,Aw30,(B12),Bw15
1553	A28,A3,Bw22,B13(B12)
1556	A1,B13,B27
1558	A10,Aw24,B7,Bw15
1559	A3,A10,Bw15,B27
1560	A2,3,B7,B18
1561	A10,Aw24,Bw35,(B12)
1562	A3,A10,B13,B18
1565	A28,Aw24,B7,Bw35
1566	A1,A28,B13,Bw35
1655	A1,A2,B13,Bw17(B23)(Bw40)
1685	A1,Aw32,B18,B27
1712	Aw32,A10,Bw15,B14,(A3)
1715	A2,A9,Bw40,(B7)
1716	A10,Bw15
1726	A28,Aw30,B5,Bw16
1775	Aw30,(Bw40),B13,B5
1779	A2,10,(B18)(Bw35)
1785	A2,A29,B12,B16
1793	Aw24,B18,B13
1805	A1,A9,B18,(B13)
1806	A1,A2,Bw17,B18,(Bw22)
1807	A1,A2,Bw35,B13
1808	A2,A10,B12,B27
1810	A1,A10,B7,B13
1814	(A1),A2,A11,B8,Bw40
1815	A2,A9,B7,B13(B12)
1817	A10-Bw35/A2-Bw21
1819	A10-Bw35/Aw24-B13
1821	A2-Bw21/A1-Bw22
1823	A10-Bw35/A28-B14
1825	A2,A3,Bw22
1827	A11-B12/A2(A28)-Bw16
1836	A28-Bw16/A3-Bw35
1838	A28(A2),Aw30,B12(B13),Bw35
1845	Aw32-Bw22/A2(A28)-Bw16
1847	A11-B12/Aw32-Bw22
1853	Aw32,w24,Bw40,B12
1855	Aw32(A1),B12,B27
1857	A9,A11,B7,Bw15
1861	A3,A2;B27,Bw22(Bw40)
1867	A2,Aw30,B12,Bw22
1868	A28,A10,Bw15,Bw16
1883	A28,Bw35
1884	A29,Aw31,33,B12,Bw15
1885	A2,A3,Bw35,Bw22
1899	A2,A11,B18,B5(Bw35)
1900	A2,A28,B18,B5(Bw35)
1901	A1(A9),A11,B8,Bw35
1902	A2,A11,B13

GM #	HL-A	GM #	HL-A
1905	A11,Bw35		
1913	A2,Aw31,B18,B13		
1930	A2,A29,B12,Bw35		

APPENDIX E

NSF CELL CULTURE CENTERS

The Cell Culture Centers at the Massachusetts Institute of Technology and the University of Alabama at Birmingham are accepting applications for large scale cell and virus production required for highly meritorious research projects. These centers established by the National Science Foundation are intended to serve as a facility and research resource for scientists throughout the country.

The MIT Center is headed by Dr. Phillips W. Robbins of MIT and Dr. Richard L. Davidson of Harvard Medical School. This facility is designed for large scale monolayer and suspension cultures and has experience with a large number of different cell lines and viruses and also with diploid human fibroblasts. The University of Alabama center is headed by J. Claude Bennett of the Department of Microbiology and Ronald T. Acton of the Diabetes Research and Training Center. This facility is designed for large scale suspension cultures with a capability of producing one kilogram or more of cells per week. Their experience to date is is primarily with lymphoblastoid cell lines. The purpose of these centers is to produce cells and viruses on a large scale in order to allow scientists to conduct novel and important experiments in basic biological research that could not be accomplished with the materials and resources in the investigators' own laboratory. Approval of applications is on

a competitive basis, based on the merit of the research project as evaluated by Centers' Steering Committees.

Examples of recent activities at the Centers are:

1. Production of 100 mg Sindbis Virus propagated in 200 roller bottles of cells for use in x-ray structural studies.

2. Production of 750 roller bottles of SV40 transformed BALB 3T3 cells for purification and characterization of T antigen.

3. Growth of 300 liters of mouse leukemia cells for isolation and structural analysis of a specific lysine transfer RNA implicated in the control of cell division.

4. Growth of 3.1×10^{12} BW 5147 cells for the isolation and structural analysis of H-2k and Thy-1 alloantigens.

Following approval of an application, the investigator sends a stock of cells or virus to the center. The stock is then grown to the requested amount, under conditions of careful handling and rigorous screening for contaminants (including mycoplasma), and prepared according to the needs of the investigator. Investigators will be asked to pay minimal costs to cover media and other supplies.

The procedure for applying involves submitting a letter specifying the amount of cell or virus required along with a brief description of the relevant research project and available supporting material. Applications or inquiries should be addressed to:

Mr. Donald Giard
Cell Culture Center
E17-321
Massachusetts Institute of Technology
77 Massachusetts Avenue
Cambridge, MA 02139

OR

Dr. Ronald T. Acton
Diabetes Research and Education Hospital
University of Alabama Hospitals and Clinics
1808 7th Avenue South
Birmingham, AL 35294

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